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With declaration under Article 17(2)(a): without abstract; title not checked by the International Searching Authority.

(54) Title: PROBES USED FOR GENETIC PROFILING

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PATENT COOPERATION TREATY

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DECLARATION OF NON-ESTABLISHMENT OF INTERNATIONAL SEARCH REPORT

(PCT Article 17(2)(a), Rules 13ter.1(c) and Rule 39)

Applicant's or agent's file reference	IMPORTANT DECLARATION	Date of mailing *(day/month/year)*
39793/JMD		12/01/2000
International application No.	International filing date *(day/month/year)*	(Earliest) Priority date *(day/month/year)*
PCT/GB 99/01779	04/06/1999	06/06/1998
International Patent Classification (IPC) or both national classification and IPC	C12Q1/68 C07K16/18	
Applicant		
GENOSTIC PHARMA LIMITED et al.		

This International Searching Authority hereby declares, according to Article 17(2)(a), that **no international search report will be established** on the international application for the reasons indicated below

1. ☐ The subject matter of the international application relates to:
   - a. ☐ scientific theories.
   - b. ☐ mathematical theories
   - c. ☐ plant varieties.
   - d. ☐ animal varieties.
   - e. ☐ essentially biological processes for the production of plants and animals, other than microbiological processes and the products of such processes.
   - f. ☐ schemes, rules or methods of doing business.
   - g. ☐ schemes, rules or methods of performing purely mental acts.
   - h. ☐ schemes, rules or methods of playing games.
   - i. ☐ methods for treatment of the human body by surgery or therapy.
   - j. ☐ methods for treatment of the animal body by surgery or therapy.
   - k. ☐ diagnostic methods practised on the human or animal body.
   - l. ☐ mere presentations of information.
   - m. ☐ computer programs for which this International Searching Authority is not equipped to search prior art.

2. ☐ The failure of the following parts of the international application to comply with prescribed requirements prevents a meaningful search from being carried out:
   ☐ the description ☐ the claims ☐ the drawings

3. ☒ The failure of the nucleotide and/or amino acid sequence listing to comply with the standard provided for in Annex C of the Administrative Instructions prevents a meaningful search from being carried out:
   - ☒ the written form has not been furnished or does not comply with the standard.
   - ☒ the computer readable form has not been furnished or does not comply with the standard.

4. Further comments: see FURTHER INFORMATION sheet PCT/ISA/203

Name and mailing address of the International Searching Authority	Authorized officer
European Patent Office, P.B. 5818 Patentlaan 2 NL-2280 HV Rijswijk Tel. (+31-70) 340-2040, Tx. 31 651 epo nl, Fax: (+31-70) 340-3016	Barbara Klaver

In view of the large number of alternative solutions to the obvious desirable objective of the provision ofdisease related nucleic acid and antibody probes, and in view of the wording of the claims as filed, it impossible to determine the matter for which protection is sought. Accordingly, the present application fails to comply with the requirement of Article 6 PCT, first sentence (see also Rule 6.1(a) PCT) and fails to comply with the requirements for clarity and conciseness of Article 6 PCT, second sentence.

Moreover, the present claims relate to an extremely large number of possible undefined probes for which no technical features are provided. The claims cover all probes having the characteristic of being disease related, whereas the application provides no support at all within the meaning of Article 6 PCT and/or disclosure within the meaning of Article 5 PCT for such probes. In the present case, the claims so lack support, and the application so lacks disclosure, that a meaningful search is impossible. Independent of the above reasoning, the claims also lack clarity (Article 6 PCT). An attempt is made to define the probes by reference to a result to be achieved. Again, this lack of clarity in the present case is such so as to render a meaningful search impossible.

The sequence listing as present in the description does not comply with WIPO Standard ST 25 prescribed in the administrative instructions under Rule 5.2. Thus, the sequences have not been provided either on paper or machine readable form in accordance with the said instructions, and the Applicant has not remedied the disclosed deficiences within the time limit fixed in the invitation to Rule 13ter.1a.

The applicant's attention is drawn to the fact that claims, or parts of claims, relating to inventions in respect of which no international search report has been established need not be the subject of an international preliminary examination (Rule 66.1(e) PCT). The applicant is advised that the EPO policy when acting as an International Preliminary Examining Authority is normally not to carry out a preliminary examination on matter which has not been searched. This is the case irrespective of whether or not the claims are amended following receipt of the search report or during any Chapter II procedure.

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(54) Title: PROBES USED FOR GENETIC PROFILING

(57) Abstract

There is considerable evidence that significant factor underlying the individual variability in response to disease, therapy and prognosis lies in a person's genetic make-up. There have been numerous examples relating that polymorphisms within a given gene can alter the functionality of the protein encoded by that gene thus leading to a variable physiological response. In order to bring about the integration of genomics into medical practice and enable design and building of a technology platfom which will enable the everyday practice of molecular medicine a way must be invented for the DNA sequence data to be aligned with the identification of genes central to the induction, development, progression and outcome of disease or physiological states of interest. According to the invention, the number of genes and their configurations (mutations and polymorphisms) needed to be identified in order to provide critical clinical information concerning individual prognosis is considerably less than the 100.000 thought to comprise the human genome. The identification of the identity of the core group of genes enables the invention of a design for genetic profiling technologies.

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PROBES USED FOR GENETIC PROFILING

People vary enormously in their response to disease and the also in their response to therapeutic interventions aimed at ameliorating the disease process and progression. However, the provision of medical care and medical management is centered around observations and protocols developed in clinical trials on groups or cohorts of patients. This group data is used to derive a standardised method of treatment which is subsequently applied on an individual basis (e.g. the comment that drugs are often prescribed on the basis that everyone is a 70kg white male).

It is standard practice for clinicians to prescribe the same starting dose of a particular drug for a given indication and then adjust the treatment regimen by monitoring the progress of the disease and therapeutic response in individual patients. Observation of *actual* therapeutic outcome following these adjustments to patient's therapy provides the basis for determining a prognosis for the disease and developing a clinical management plan for patient care (e.g. see Fig 1, algorithm for management of schizophrenia, from Fig 1 Taylor and Kerwin 1997, Fig 2 algorithm for treatment of depression from Fig 1 Pathare and Paton 1997) and treatment algorithms published by the National Cancer Institute).

The standard practice of clinical management has its disadvantages. In particular it is retro-active in that changes to patient management will occur following the emergence of therapeutic failures, adverse events or other difficulties in undertaking the therapeutic regime (Lazarou et al 1998).

There is considerable evidence that a significant factor underlying this individual variability in response to disease, therapy and prognosis lies in a person's genetic make-up. There have been numerous examples relating that polymorphisms within a given gene can alter the functionality of the protein encoded by that gene thus leading to a variable physiological response (see Marshall 1997a and b for reviews).

Gene sequence variations that are present at a frequency of less than 1% in the population are arbitrarily designated as mutations whilst those at a higher frequency are known as polymorphisms (Schafer and Hawkins 1998).

DNA variants leading to monogenic diseases (e.g. presenilin mutations causing Alzheimer's disease, BRCA mutations causing breast cancer) are usually rare in a population due to the process of natural selection. However, variants of genes involved in, or contributing to, polygenic diseases do not act alone to produce the phenotype. As such selection against them occurs only when they are in the appropriate condition to cause the disease, as a result of this differential selection pressure they the individual variants can exist at quite high frequencies within a population.

Alteration of a single gene may not by itself be detrimental, but in combination with certain variants of other genes, may contribute to a disease phenotype (e.g. el-Zein et al, 1997, observed that the inheritance of a particular combination of metabolising genes is strongly associated with lung cancer). The interaction of the relevant variant genes may be enough to cause a disease phenotype or spectrum of phenotypes, but in

many cases other kinds of factors will also influence the course of events (e.g. interaction of ApoE genotype and head injury in Alzheimer's disease Nicholl et al 1996).

The identification of modifier genes that influence the penetrance and expressivity of these risk alleles will be key variables in assessing individual risk profiles. It is likely that the combination of and interaction between small discrete genetic influences on a disease state represent the single largest explanation for the phenotypic variation seen in medicine.

This opens the possibility that the identification of the genes associated with disease and an understanding of how these genes interact with the environment, can lead to better prediction of the outcome of both the disease and the therapeutic process. This in turn would allow the tailoring of resources and therapy to meet the likely requirements of the individual patient (Marshall 1997a). The net result should be improved clinical management, identification of the potential for prevention, the reduction of the burden of disability and, ultimately, improved quality of life for the individual (Poste 1998).

As a result of the appreciation of the contribution of genetic variation to medicine, considerable effort has been made to determine how individual genetic variations affect overall health (including predisposition to disease) and once disease is manifest, the likely patterns of progression, responsiveness to treatment and overall prognosis.

In a quest to understand and plot the limits of genetic variation in humans the Human Genome Project was launched in 1990 with a mission to sequence the code of all 100,000 or so human genes by 2002.

As a result of the Human Genome project not only is the mapping and sequencing of the human genome becoming well understood but also the degree of variability in gene sequence between individuals is being documented (Lander 1996). The average difference between individuals appears to be around 0.3% which equates roughly to a difference in one base pair every 500-1000 base pairs of sequence. The variations are known as polymorphisms and such polymorphic variation is thought underlie much of the clinical variability observed in patients with disease and in their response to therapy.

The resultant explosion of genetic sequence information has lead to the emerging sciences of genomics and proteomics. Within the disciplines technologies have evolved (e.g. polymerase chain reaction, single strand conformational polymorphism etc) which allow us to read individual sequence data and detect and identify polymorphic variation in individuals, in disease states and in different ethnic groups (Griffin et al 1997, Little et al 1997).

As a result of such studies individual genes have been identified which indicate a predisposition to disease or a susceptibility to adverse drug responses (e.g. presenilin gene mutations and development of Alzheimer's disease, BRCA gene mutation and development of breast cancer, ACE polymorphisms and early onset heart disease, cytochrome P450 polymorphisms and drug metabolism).

However, such studies have been completed as academic exercises in scientific discovery and involve individual genes and large groups of patients.

Usually a particular individual response to disease or therapy is likely to result from a complex interaction between multiple genes, discrete environmental factors and the particular therapeutic approach offered (for example see algorithms in Figs. 1 and 2).

As a result, despite the many publications concerning the theoretical or potential applications of genomics to medicine (e.g. Marshall 1997a and b, Poste 1998, Crooke 1998), progress in implementing these approaches on a practical level has been exceedingly slow. In particular, little progress has been made in the understanding of or the ability to prognose individual response to particular disease states or therapeutic regimes (Poste 1998).

In part this has been related to the types of technology available for such studies (Marshall and Hodgson 1998). Such techniques as MALDI-TOF (Griffin et al 1997), sequencing (Dramanac et al 1998) and molecular beacons (Tyagi et al 1998) are complex and relatively slow and require the availability of specialised laboratories and highly trained personnel.

In recent reviews of the field it has been stated that:

- 'within next 10 years when not only all genes (will have been) identified but all common intragenic variation also' (Lander 1996).

- the 'assembly of comprehensive clinical databanks and their use for large-scale genetic association studies to define robust disease-gene risk correlations' constitutes a significant technological challenge (Poste 1998).

- 'if all human DNA variants were known this set would include all functional polymorphisms and if they could be analysed in all individuals comparison of phenotypes and correlation with genotype might make possible the assignment of function to every gene that predisposes to disease of any kind, and also to non-clinical phenotypes including behavioural traits. **The sheer task of this is overwhelming and may never be practical'** (Shafer and Hawkins 1998).

On the basis of the current state of the art it seems clear that translating the colossal investment in the human genome project into a means of revolutionising healthcare management requires both substantial creativity in the harnessing of technologies and considerable technical invention before its promise of can be realised.

For the realisation of the promised revolution in medicine two key factors require consideration;

- The human genome is made up of some 100,000 separate genes.

- Not all genes are of equal biological importance as regards the physiological functioning of humans.

The first issue, that of reading and tracking the volume of information encapsulated in the human genome by the sequence of 100,000 genes and their mutations and polymorphic variations, is beginning to be addressed by emergent technologies such as DNAchips, MALDI-TOF MS (Marshall and Hodgson 1998 see Table 1) and PEDIAT-type technologies (Fox 1998).

Table 1. The main features of some hybridization array formats currently available (Marshall & Hodgson 1998)

Company	Arraying method	Hybridization step	Readout	Main focus
Affymetrix (Santa Clara, CA)	On-chip photolithographic synthesis of ~20-25-mer oligos onto silicon wafers, which are diced in 1.24 cm^2 or 5.25 cm^2 chips	10,000-260,000 oligo features probed with labelled 30-40 nucleotide fragments of sample cDNA or antisense RNA	Fluorescence	Expression profiling, polymorphism analysis, and diagnosis
Brax (Cambridge, UK)	Short synthetic oligo, synthesized off chip	1,000 oligos on a "universal chip" probed with tagged nucleic acids	Mass spectrometry	Diagnostics, expression profiling, novel gene identification
Hyseq (Sunnyvale, CA)	500-2000 nt DNA samples printed onto 0.6 cm^2 (HyGnostics) or ~18 cm^2 (Gene Discovery) membranes	64 sample cDNA spots probed with 8,000 7-mer oligos (HyGnostics) or ≤55,000 sample cDNA spots probed with 300 7-mer oligos (Gene Discovery)	Radioisotope	Expression profiling, novel gene identification, and large-scale sequencing (Gene Discovery array), polymorphism analysis and diagnostics (HyGnostics/ HyChip arrays), and large-sample sequencing (HyChip array)
	Prefabricated 5-mer oligos printed as 1.15 cm^2 arrays onto glass (HyChip)	Universal 1024 oligo spots probed 10 kb sample cDNAs, labelled 5-mer oligos and ligase	Fluorescence	
Incyte Pharmaceuticals (Palo Alto, CA)	Piezoelectric printing for spotting PCR fragments and on-chip synthesis of oligos	≤ (eventually 10,000) oligo/PCR fragment spots probed with labelled RNA	Fluorescence and Radioisotope	Expression profiling Polymorphism analysis, Diagnostics
Molecular Dynamics (Sunnyvale, CA)	500-5000 nt cDNAs printed by pen onto ~10 cm^2 on glass slide	~10,000 cDNA spots probed with 200-400 nt labelled sample cDNAs	Fluorescence	Expression profiling and novel gene identification
Nanogen (San Diego, CA)	Prefabricated ~20 mer oligos, captured onto electroactive spots on silicon wafer, which are diced. Into ≤ 1 cm^2 chips	25, 64, 100, 400 (and eventually 10,000) oligo spots polarized to enhance hybridization to 200-400 nt labelled sample cDNAs	Fluorescence	Diagnostics and short tandem repeat identification
Protogene Laboratories (Palo Alto, CA)	On-chip synthesis of 40-50-mer oligos onto 9 cm^2 glass chip via printing to a surface-tension array	≤8,000 oligo spots probed with 200-400 nt labelled sample nucleic acids	Fluorescence	Expression profiling, and polymorphism analysis
Sequenom (Hamburg, Germany and San Diego, CA)	Off-set printing of array, around 20-25-mer	250 locations per SpectroChip interrogated by laser desorbtion and mass spectrometry	Mass spectrometry	Novel gene identification, candidate gene validation, diagnostics, and mapping
Synteni (Fremont, CA)	500-5000 nt cDNAs printed by tip onto ~4 cm^2 glass chip	≤10,000 cDNA spots probed with 200-400 nt labelled sample cDNAs	Fluorescence	Expression profiling and novel gene identification

The German Cancer Institute (Heidelberg, Germany)	Prototypic DNA macrochip with on-chip synthesis of probes using f-moc or t-boc chemistry	Around 1000 spots on a 8x12 cm chip	Fluorescence/ mass spectrometry	Expression profiling and diagnostics

These new technologies mark a significant advance in the potential application of genomic information to the problems of biology and human health. The reason for this is their capability of determining or confirming a large volume of DNA sequence data very quickly at the individual level. In this way they open the door to the application of genomic information to the individual patient.

These technologies are also evolving quickly according to Moore's Law (which posits that computer chips' power doubles every 18 months). For instance, three years ago the genechips made by leading companies held some 20,000 DNA probes. Currently genechips with 65,000 probes are available, and a chip with 400,000 probes has recently been produced (Marshall and Hodgson 1998). Applications for such technologies have included sequencing, diagnostics (mutation detection in the BRCA1 gene for cancer), gene discovery, gene expression profiling and gene mapping (Marshall and Hodgson 1998).

However despite their value as research and diagnostic tools, the genechips in existence are utilized largely as research tools (Marshall and Hodgson 1998). They have not been used as a tool for the express purpose of improving healthcare management by enabling the process of clinical prognosis and facilitating the generation of health risk profiles.

The reason for this is the failure to conceive of or invent an appropriate design which identifies the critical core of genes which are the most important in terms of human function. The genetic variability in this group of genes is the most important contributor to the variation in clinical and physiological phenotypes. Not all genes are equally important in the normal physiological functioning of the human body nor in the induction, development or progression of diseases or physiological states. In a given disease, as few as 5-10 genes in different configurations may be of seminal importance in determining the vast bulk of inter-individual variability to disease and therapeutic approaches (Drews 1997, Goodman and Gillman 1996).

As such, a device capable of delivering information on 10,000 genes may leave its user in grave danger of information overload and render him/her unable to identify and abstract the critical information required to enhance patient management or healthcare.

As a result, the translation of such technologies in genechip devices from research tools into healthcare management tools is severely limited (Marshall and Hodgson 1998, Poste 1998, Schafer and Hawkins 1997).

In an effort to overcome this difficulty a consortium of academic and industrial groups (SNP Consortium) has been formed to try and identify the important disease related variants of human genes. The technologies to be used are the generation and assembly

of a SNP map spanning the whole human genome and its application to linkage studies.

However, this approach is still in its infancy and is widely held to face considerable technical hurdles in the robust statistical analysis of huge datasets.

In order to bring about the integration of genomics into medical practice and enable design and building of a technology platform which will enable the everyday practice of molecular medicine a way must be invented for the DNA sequence data to be aligned with the identification of genes central to the induction, development, progression and outcome of disease or physiological states of interest:

Practitioners of molecular healthcare need to be able to;

- Identify the presence or absence of a selected group of genes and polymorphic variants central to the induction, development progression and outcome of disease or physiological states
- Focus on polymorphisms that lie within the coding or regulatory regions of the gene and are likely to result in altered structure or expression of the protein.
- Utilise the data on the core group of genes in order to generate guidelines and guidance for the healthcare management of patients or persons.

The invention described herein identifies the core group of genes required for the design development and manufacture of such a valuable aid to clinical management of the patient and general healthcare management.

According to the invention, the number of genes and their configurations (mutations and polymorphisms) needed to be identified in order to provide critical clinical information concerning individual prognosis is considerably less than the 100,000 thought to comprise the human genome.

The identification of the identity of the core group of genes enables the invention of a design for genetic profiling technologies which comprises of the identification of the core group of genes and their sequence variants required to provide a broad base of clinical prognostic information - 'genostics'.

By careful and lengthy research of the literature, tabulation of data, cross referencing of studies and conduction of a variety of experiments we have identified the core group of genes, which, if assessed for the presence of their functional variants, will enable an enhanced prognosis for an individual patient and form the basis for converting genetic profiling technologies from research tools into universal tools for health management.

Identification of the core group of genes and their functional variants also allows for said technologies to be utilised in generating individual health-risk profiles and profiling the health-risks of the population at large. The determination and identification of sequence data required to identify the important functional variants is readily accomplished by those skilled in the practice of the relevant arts.

The invention does not provide a method for treatment as such. Nor does it provide a direct method of diagnosis of illness or health risk as such. Information obtainable using the invention can be used by a medical practitioner to tailor resources and therapy to meet the likely requirements of individual patients and selected populations of patients. For example in a complex regime or clinical management plan (as seen for example in Fig. 1 and 2) the invention allows the better prediction of the outcome of both the disease and the chosen therapeutic process.

The enablement of the invention and the generation of the information required for the design of 'genostics' requires:

1. Identification of sequence data (Example 1).
2. Assessment of the type and significance of sequence variation in the core group of genes (Examples 2,3,4).
3. Identification of likely genetic variation/disease relationships (Example 5 and 5a).
4. Means of identifying and detecting additional polymorphisms in the core group of genes (Example 6).
5. A practical approach to data analysis to generate information on prognosis(Example 7).
6. An illustration of how clinical management of a patient can be enhanced by utilising genetic profiling approaches (Example 8 and 9).

EXAMPLE 1

Gene sequence data is readily available in the public domain.

For the design of the GENOSTIC genechip device, gene sequence data can be retrieved, by persons skilled in the art, by searching the following public databases:

Website	Address	Description
DbEST	http://www.ncbi.nlm.nih.gov/dbEST	Database of expressed sequence tags
EBI/EMBL	http://www.ebi.ac.uk/mutations/	Mutations
EBI: The European Bioinformatics Institute, Hinxton, UK	http://www.ebi.ac.uk/ebi_home.html	Nucleotide Sequence Database
EMBL	http://www.ebi.ac.uk/queries/queries.html	Nucleotide Sequence Database
GDB: The Genome Database, Infobiogen European Node, FRANCE	http://www.gdb.org/gdb/gdbtop.html	Human Genome Database

GeneCards	http://bioinformatics.weizmann.ac.il/cards/index.html	**GeneCards** is a database of human genes, their products and their involvement in diseases.
GeneClinics	http://www.geneclinics.org/	***GeneClinics (formerly Genline)*** is a knowledge base of expert-authored, up-to-date information relating genetic testing to the diagnosis, management, and counseling of individuals and families with inherited disorders.
Genethon	http://www.genethon.fr/genethon_en.html	The Human Genome Research Centre.
GSDB: Genome Sequence database	http://www.ncgr.org/	A collection of DNA sequence data and related information.
HGP: Human Genome Project Information	http://www.ornl.gov/TechResources/Human_Genome/home.html	Useful background & links.
Human Gene Mutation Database	http://www.uwcm.ac.uk/uwcm/mg/search	Mutations
NCBI	http://www.ncbi.nlm.nih.gov/	KEY SITE. Nucleotide Sequence retrieval start point.
OMIM: Online Mendelian Inheritance in Man	http://www.ncbi.nlm.nih.gov/Omim/	This database is a catalog of human genes and genetic disorders.
PubMed	http://www.ncbi.nlm.nih.gov/PubMed/	PubMed accesses MEDLINE medica literature database and links to full-text journals. It is also the literature component of the Entrez retrieval system for molecular biology information.
Research Tools (Science - NCBI)	http://www.ncbi.nlm.nih.gov/SCIENCE96/ResTools.html	A Gene Map of the Human Genome.
RHdb: Radiation Hybrid Database, Hinxton, UK	http://www.ebi.ac.uk/RHdb	Radiation Hybrid Database.
Stanford Human Genome Centre	http://www.shgc.stanford.edu/	Sequence database.
HUGO: The Human Genome Organisation	http://www.gene.ucl.ac.uk/hugo	HUGO is the international organisation of scientists involved in the Human Genome Project.
TIGR: The Institute for Genomic Research	http://www.tigr.org/	Genomic databases.
The National Human Genome Research Institute	http://www.nhgri.nih.gov/	Access to sequence databases
The Whitehead Institute Center for Genome	http://www.genome.wi.mit.edu/	Genome map and sequence information.

Research		
Unigene: Unique Human Gene Sequence Collection. (NCBI)	http://www.ncbi.nlm.nih.gov/UniGene/index.html	UniGene is a system for automatically partitioning GenBank sequences into a non-redundant set of gene-oriented clusters. Each UniGene cluster contains sequences that represent a unique gene, as well as related information such as the tissue types in which the gene has been expressed and map location.
University of Oklahoma	http://dna1.chem.ou.edu/index.html	Genomic databases
WEHI, Melbourne, Aus	http://wehih.wehi.edu.au/srs/srsc/	Sequence Retrieval System

Genes coding for proteins known to play a key role in organ function or disease are designated 'candidate genostic genes'. Variations within the gene structure may alter the regulatory or structural integrity of the gene product leading to enhancement or reduction in the specific function (e.g. receptor binding, enzyme activity). The exact role that a candidate gene plays in disease, prognosis and healthcare management can be fully ascertained by assessing the effects of variation in gene structure in particular patient groups, populations or individuals (see examples 2,3 and 4).

EXAMPLE 2 -Candidate Genostic Genes

Human Neuronal Nitric Oxide Synthetase
Gene Map Locus: 12q24.2q24.31(OMIM Ref. 163731).

One candidate 'genostic' gene is the gene encoding nitric oxide synthetase (NOS-1).

The enzymes responsible for NO synthesis in man constitute a family with at least three distinct isoforms: inducible, endothelial, and neuronal. Neuronal NO synthetase (NOS-1) is localised to human chromosome 12, and participates in diverse biologic processes including neurotransmission, the regulation of body fluid homeostasis, neuroendocrine physiology, control of smooth muscle motility, sexual function and monocyte biology.

Burnett et al. (1992) localized NO synthase to rat penile neurons innervating the corpora cavernosa and to neuronal plexuses in the adventitial layer of penile arteries. They demonstrated that small doses of NO synthase inhibitors abolished electrophysiologically induced penile erections establishing that nitric oxide is a physiologic mediator of erectile function.

Kharazia et al. (1994) found that all neurons in the striatum and many in the cortex were positive for nitric oxide synthase indicating a role of NOS in brain function.

NOS1 cDNA clones contain different 5-prime terminal exons spliced to a common exon 2. Xie et al. (1995) demonstrated that the unique exons are positioned within 300 bp of each other but separated from exon 2 by an intron that is at least 20 kb long. A CpG island engulfs the downstream 5-prime terminal exon. In contrast, most of the upstream exon resides outside of this CpG island. The upstream exon includes a GT dinucleotide repeat. The expression of these 2 exons is subject to transcriptional control by separate promoters. Nitric oxide is synthesized in skeletal muscle by neuronal-type NO synthase, which is localized to sarcolemma of fast-twitch fibers. Synthesis of NO in active muscle opposes contractile force. Brenman et al. (1995) showed that NOS1 partitions with skeletal muscle membranes owing to association of enzyme with dystrophin, the protein mutated in Duchenne muscular dystrophy. The dystrophin complex interacts with an N-terminal domain of NOS1 that contains a GLGF motif. Both humans with DMD and mdx mice show a selective loss of NOS1 protein and catalytic activity from muscle membranes. NOS1-deficient mice are resistant to neural stroke damage following middle cerebral artery ligation. Nelson et al. (1995) reported a large increase in aggressive behavior and excess, inappropriate sexual behavior in NOS1 'knockout' mice. Initial observations indicated that male (but not female) NOS1-deficient mice engaged in chronic aggressive behavior.

Magee et al. (1996) used PCR to clone a novel form of neuronal NOS from rat penile RNA. This NOS cDNA was termed PnNOS for 'penile neuronal NOS.' Sequencing revealed that the PnNOS cDNA was identical to rat cerebellar neuronal NOS1 except for a 102-bp insertion in PnNOS. Repetition of RT-PCR showed PnNOS to be the only form of NOS1 expressed in rat penis, urethra, prostate, and skeletal muscle. PnNOS may be responsible for the synthesis of nitric oxide during penile erection and may be involved in control of the tone of the urethra, prostate, and bladder.

Using the available genomic sequence of neuronal NOS-1 it is possible to identifiy those parts of the gene which show variation sufficient to alter the normal functioning of the gene.

1.) Transcriptional Promoter Sequences:

Sequence mutations in the promoter region of the NOS1 gene will allow the identification of individuals with altered transcriptional regulation control.

2.) RNA Processing (Splicing) Sequences:

Characterise mutations in the intron/exon structure of the NOS1 gene to identify individuals with altered RNA splicing patterns. These results in truncated proteins or splice variants with an altered function.

3.) Messenger RNA Translation and Stability Sequences:

Sequence and characterise mutations within the repetitive sequences located in the 3' untranslated region of the NOS-1 gene. These individuals have altered translational control of their mRNA.

4.) DNA Sequences Involved in Genomic Rearrangement or Expansion:

The presence of Alu-1 repeat, which are known to cause recombination, allows one to detect gross chromosomal rearrangements. Changes in either the sequence or the genomic structure may well correlate with clinical or pathological symptoms.

102-bp insertion will also be involved in the functional variation of activity involving the urogenital tract.

5.) Coding Sequences:

Mutations and polymorphisms in the coding (exon) sequences of the NOS-1 gene will result in changes at the structural level of the protein with functional changes. Amino acid substitutions, within neuronal NOS-1, will play a role in age/brain related neuronal defects.

The specific sequences are detailed in Table 2.

TABLE 2: Summary of Genome Elements within the Neuronal Nitric Oxide Synthetase Gene.

Gene Anatomy	Key Region	Functional Elements
1. 5' Flanking Region:	GC-enriched sequences:	DNA methyltransferase foot print region CpG Island
	Promoter elements	TATA box Inverted CAAT boxes AP-2-like element CREB/ATF element c-Fos element NF-kB-like ETS-binding sites TEF-1/MCBF binding sites NRF-1 binding sites RNA Pol III site
2. Exon Coding Regions		Translation initiation exon 2 Translation termination exon 29
3. RNA Processing		Intron/exon boundaries (1-29) Cassette splicing exons 9-11
4. RNA Translation		3' Untranslated Region
5. Insertion		102bp insertion
6.Repetitive Sequences		Alu-1 family Dinucleotide repeats

These variations in the genomic structure of the human NOS 1 gene are important in controlling the physiological role of NOS in normal or disease states in humans. Alterations in the physiology of NOS have significant healthcare indications (i.e stroke, cardiac and circulatory disease, urogenital disease and dysfunction, psychiatric symptoms and musculoskeletal disorders).

In consideration with an assessment of the functional variation in other genes, identification of the pattern of NOS 1 gene variation in a patient cohort, population or individual offers a powerful practical tool for improving the management of healthcare and the prognosis of health risk.

EXAMPLE 3

Voltage-gated calcium channels
Gene map locus (OMIN Ref.601011)

Other candidate 'genostic' genes are the calcium channel subunit genes.

There are six functional subclasses of calcium channel. Voltage-dependent Ca(2+) channels not only mediate the entry of Ca(2+) ions into excitable cells but are also

involved in a variety of Ca(2+) – dependant processes, including muscle contraction, hormone or neurotransmitter release and gene expression.

Calcium Channels are multi-subunit complexes and the channel activity is directed by a pore-forming alpha-1 sub-unit. The auxillary sub-units beta, alpha-2/delta, and gamma regulate channel activity. Ca(2+) currents have been described on the basis of their biophysical and pharmacological properties and include L-, N-, T-, P-, Q-, and R- types.

P/Q type channels colocalise with a subset of docked vesicles at the synapse where they control exocytosis, demonstrated by the sensitivity of various types of neurotransmission to specific blockers of these channels. P/Q type channels are involved in CSD (cortical spreading depression – which causes the aura or visual symptoms of migraine) and release of neurotransmitters, including 5-HT (migraine patients have systemic disturbance of 5-HT metabolism).

The distinctive properties of each of the Ca(2+) channel types are primarily related to the expression of a variety of alpha-1 isoforms (Dunlap *et al.*, 1995). There are at least 6 classes of alpha-1 subunits: alpha-1A, B, C, D, E and S. They are derived from 6 genes representing members of a gene family. The alpha-1A, B and E isoforms are abundantly expressed in the neuronal tissue. The genes encoding the alpha-1A, B, and E isoforms are symbolised CACNL1A4, CACNL1A5, and CACNL1A6 respectively.

The CACNL1A4 gene was assigned to 19p13, (Diriong *et al.*, 1995). The gene was characterised by Ophoff *et al.* (1996) in preparation for a mutation search in neurological disorders that map to 19p13. They found that the gene covers 300 kb with 47 exons and reported the amino acid sequence for residues 1-2262. Sequencing of all the exons and their surroundings revealed polymorphic variations, including a (CA)n-repeat, a (CAG)n-repeat in the 3-prime-UTR, and different types of deleterious mutations in 2 neurological disorders; familial hemiplegic migraine and episodic ataxia type 2. Thus, these 2 neurological disorders are allelic channelopathies.

Calcium channels are also known to be important in regulating the function of the heart (particularly arrhythmias) and a number of drugs express their therapeutic effects by blocking myocardial Ca(2+) or prolonging the activation time of the channel (Brody, Larner and Minneman 1998). Polymorphic variation can help predict individual response to injury and disease, the symptoms and consequences of cardiovascular disease, dysfunction and damage to the system.

EXAMPLE 4

Lipoprotein lipase LPL
Gene map locus (OMIN Ref.238600)

A third example of a candidate for a 'genostic' gene is the enzyme lipoprotein lipase (LPL).

Human lipoprotein lipase is a member of a lipase gene family, which also includes the hepatic and pancreatic lipases. LPL is located on the surface of endothelial cells of

capillaries where it hydrolyses triacylglycerols of plasma lipoproteins to fatty acids and glycerol. These fatty acids are then taken up by cell and used for energy production. The enzyme plays a central role in lipid metabolism and is a candidate susceptibility gene for cardiovascular disease.

The LPL gene contains ten exons spanning 30kb and encodes a protein of 475 amino acids and has several well characterised functional domains including the APOC-II binding site, the heparin-binding clusters used to localise LPL to the endothelial wall and the domains that contribute to the active site.

Diseases that affect the metabolism and transport of lipids frequently result in abnormally high plasma triacyglycerols and or cholesterol that are often associated with coronary artery disease, artherosclerosis and/or obesity. DNA sequence variation in genes that encode many of the enzymes and proteins involved in lipid metabolism and transport (including LPL) have been identified and associated with clinically abnormal lipid profiles.

The LPL gene sequence has been shown to contain distinct sequence variations among populations, (Nickerson *et al*, 1998). Nickerson *et al* described 88 variants in a region of the LPL gene, 90% of which were single nucleotide polymorphisms (SNPs), the remaining being insertion-deletion variations. 81 variants were found in intronic regions, and 7 in the exonic sequence. Only 4 of the exonic variants altered the protein sequence.

Assessing the functional variability of the LPL gene in conjunction with the functional variabilty of other core genes will provide a tool in predicting the likelihood of developing a range of diseases including the symptoms and consequences of coronary artery disease, artherosclerosis and/or obesity.

As shown above, sequence data for genes of interest can be readily obtained. Genetic variation in specific regions of genes can also be determined. The identification of a core group of genes which have important effects on the key physiological and pathophysiological processes in human disease would form an important medical advance.

A device or detector configured and designed using this core group of genes (GENOSTIC) would have a general utility in the practice of medicine and healthcare management for:

- prognosing the course of illness
- predicting likely therapeutic response
- identifying potential adverse event profile.

EXAMPLE 5

LIST OF GENES WITH KNOWN ASSOCIATION WITH DISEASE

The following are examples of **genes with known associations with disease** which can be discerned by a careful review of the medical and biochemical literature and by experimentation. Many such genes can also be identifed by a review of publicly available databases e.g. Human Gene Mutation Database (http://www/uwcm.ac.uk/uwcm/mg/search/), OMIM Database (http://www.ncbi.nlm.nih.gov/omim) or GENECARDS (http://bioinformatics.weizmann.ac.il/cards/index.html).

Note: The tabulated genes are listed in alphabetical groups, but the numbering of genes within each group is not necessarily continuous.

A	B	C	D
1: APOA4	1: BLM	1: CRYAA	1: DPYD
2: AAC2	2: BCKDHA	2: CRYBB2	2: DIAPH1
3: AD2	3: BTD	3: CHM	3: DMD
4: AGA	4: BPGM	4: C2	4: DPYS
5: APOA1	5: BRCA2	5: C5	5: DFN1
6: ALAS2	6: BRCA1	6: C9	6: DKC1
7: ALB	7: BCP	7: C3	7: DLD
8: APT1	8: BLMH	8: C7	8: DFNA5
9: APOA2	9: BCKDHB	9: CTNS	9: DTD
10: APOH	10: BCHE	10: C1QA	10: DCX
11: AMELX	12: BTK	11: C1QB	11: DYT1
12: APT1LG1	13: BARD1	12: CNGA3	12: DMPK
13: A2M	18: BSEP	13: C1QG	13: DRD4
14: APBB1		14: CPO	14: DDB2
15: AGXT		15: CDH1	15: DIAPH2
16: AGTR1		16: C4A	16: dgcr5
17: ALDH2		17: C4B	17: DRD2
18: ARG1		18: C6	18: DES
19: ALD		19: C8B	19: DBT
20: AGT		20: CACT	20: DCP1
21: ACHE		21: chit	24: DYSF
22: ADSL		22: CLCN1	27: DRA
23: ADRB3		23: CFTR	29: DLX3
24: atpsk2		24: COL10A1	31: DRPLA
25: ATM		25: CYP1A1	38: DIA1
26: ASPA		26: CLCNKB	39: DHAPAT
27: ACTC		27: CD3G	
28: ADRB2		28: CACNA1F	
29: AIRE		29: CPS1	
30: AZF1		30: CRX	
31: AT3		31: CYBA	
32: ABO		32: CKN1	
33: ABCR		33: CST3	
34: AACT		34: CNGA1	
36: ANK1		35: CETP	
37: ALAD		36: CAT	
38: APOE		37: CTSK	
39: APP		38: CYBB	
40: APOC3		40: CSX	

E	F	G	H
1: EPOR	1: FUCA1	1: GM2A	2: HD
2: EPB41	2: FRDA	2: GYPC	3: HK1
3: EMX2	3: FGB	3: GALT	5: HBG2
4: EXT2	4: FH	4: GLB1	6: HSD3B2
5: EMD	5: FGG	5: GALE	7: HBG1
6: ED1	6: FMR2	6: GAMT	9: HFE
7: ESR	7: FGFR1	7: GYPA	10: HTN3
8: EXT1	8: FGA	8: GPI	11: HOXA13
9: EPHX1	9: F10	9: GPC3	12: HR
10: EPX-PEN	10: FUT6	10: GLI3	13: HBA1
11: EDNRB	11: FKHL15	11: GCDH	14: HMGCL
12: EPM2A	12: FRAXF	12: GAA	15: HBD
13: EDN3	13: FBP1	13: G6PC	16: HTR2C
14: ETFA	14: F11	14: GBA	18: HP
15: ETFB	15: F12	15: GALK1	19: HSD11B2
16: ENG	16: FCGR1A	16: GBE1	20: HK2
17: EPB42	17: FBN2	17: GLS	21: HPS
18: ETFDH	18: FAH	18: G6PT1	23: HGD
19: EFE2	19: FSHR	19: GLUD1	25: HBA2
20: ERCC5	20: F13B	20: GRL	26: HCF2
22: ERCC4	21: FMO3	21: GSS	27: HRG
23: ELN	22: FUT3	22: GK	28: HOXD13
24: EYA1	23: F13A1	23: GP1BB	29: HEXB
25: ERCC6	24: FANCA	24: GSN	32: HLCS
26: ERCC3	25: F7	25: GCGR	33: HPRT1
27: EGR2	26: FTL	26: GLRA1	34: HBB
28: ERCC2	27: F5	27: GH1	35: HTR1A
	28: FUT2	28: G6PD	36: HSD17B1
	29: FMR1	29: GYS2	37: HSD17B3
	30: FCMD	30: GHRHR	40: HSD17B4
	31: FGDY	31: GH2	
	32: FANCC	32: GCP	
	33: FCGR2A	33: GALC	
	34: FGFR3	34: GP9	
	35: FECH	35: GNRHR	
	36: FSHB	36: GIPR	
	37: F8C	37: GSTT1	
	38: FBN1	38: GLA	
	39: FABP2	39: GRPR	
	40: F9	40: GPD2	

I	J	K	L
1: IL2RA	1: JAG1	1: KRT9	1: LPL
2: IVD	2: JAK3	2: KCNQ3	2: LIPC
4: IFNGR1		3: KRT1	3: LOR
5: IL2RG		4: KNG	4: LDLR
6: IFNGR2		5: KRT16	5: LYZ
7: IGHG2		6: KRT18	6: LIG1
9: INSR		7: KRT6A	7: LDHA
10: IDUA		8: KRT6B	8: LDHB
11: IL4R		9: KRT3	9: LQT2
12: ITGA7		10: KHK	10: LEPR
13: ITGA2B		11: KRTHB1	11: LHCGR
14: IGKV		12: KEL	12: LEP
15: IAPP		13: KRTHB6	13: LHB
16: IPF1		14: KAL1	14: LIPA
17: INS		15: KRT4	15: LAMA3
18: IGF1		16: KRT13	16: L1CAM
19: IGHM		17: KRT2A	17: LAMC2
20: ITGA6		18: KRT12	19: LCAT
21: IRS1		19: KRT5	20: LAMA2
22: ICAM1		20: KRT14	21: LMX1B
23: ITGB3		21: KRT10	22: LTBP2
24: ITGB4		22: KRT17	23: LMAN1
25: IDS		23: KCNQ2	26: LAMB3
28: ITGB2		24: KCNQ1	
		26: KCNJ1	
		28: KCNJ11	
		30: KCNA1	
		32: KIT	
		36: KCNE1	

M	N	O	P
1: MTM1	1: NME1	1: OA1	1: PROP1
2: MUT	2: NF1	2: OCA2	2: PLP
3: MTR	3: NBS1	3: OCRL	3: PRPS1
4: MLH1	4: NPHP1	4: OXCT	4: PEPD
5: MMP3	5: NF2	5: OPHN1	5: PCCB
6: MVK	6: NCF1	6: OTC	6: PCCA
7: MANBA	7: NDP	7: OAT	7: PCSK1
8: MTRR	8: NCF2	8: COL1A2	8: PAH
9: MANB	9: NP		9: POU1F1
10: MPO	10: NEU		10: PPOX
11: MYO5A	11: NTF3		11: PRKCG
12: MYH7	12: NOTCH3		12: PXMP1
13: MAOA	13: NRTN		13: PPGB
14: MYOC	14: CHRNA4		14: PRB3
15: MADH4	15: NPC1		15: PRB1
16: MEFV	16: NAGA		16: PRB4
17: MAT1A	17: NEFH		17: PMP22
18: MEN1	18: NTRK1		18: PABP2
19: MOCS1	19: NAIP		19: PEX7
20: mocs1b	20: NDUFS4		20: PDDR
21: MLR	21: NOS3		21: PAFAH2
22: MSH2	23: NODAL		22: PARK2
23: MSX2	25: NAGLU		23: PLG
25: MPI			24: PPARG
26: MC4R			25: PON2
28: MDCR			26: PROC
29: MBL			27: PROS1
30: MJD			28: PDE6A
31: MC2R			29: PXMP3
32: MYL2			30: PPP1R3
33: MC1R			31: PON1
34: MYO15			32: PEX1
35: MAPT			33: PC
36: MPZ			34: PENK
37: MID1			35: PXR1
38: MSX1			36: PGK1
39: MGAT2			37: PTH
40: MTHFR			38: PDE6B
			39: PSEN2
			40: PKD2

Q	R	S	T
1: QDPR	1: RHO	1: SSA1	1: TAT
	2: RP2	2: SOD1	2: THBD
	3: RLBP1	3: COL2A1	3: TNNT2
	4: RHD	4: SDH2	4: TF
	5: RB1	5: SGSH	5: TBG
	6: ROM1	6: SLC5A5	6: TSC1
	7: RP3	7: SLC12A3	7: TCN2
	8: RHCE	8: SDH1	8: TPI1
	9: RHAG	9: SUOX	9: TPM1
	10: RHOK	10: STS	10: TBXA2R
	12: rfxank	11: ssadh	11: TPMT
	13: REN	12: SALL1	12: TYR
	14: RYR1	13: SHOX	13: TGM1
	15: RS1	14: SLC12A1	14: TTR
	16: RDS	15: SLC2A2	15: TSC2
	17: RFC2	16: SNRPN	16: TG
	18: RCP	17: SPTB	17: TTPA
	21: RFXAP	18: SCA2	18: TCOF1
	22: RAG2	19: SMN1	19: TULP1
	23: RPS6KA3	20: STK11	20: TNF
	24: RPE65	21: SPTA1	21: THPO
	25: RFX5	23: SH2D1A	22: TCF2
	26: RAG1	24: SCNN1B	23: TPO
		25: SI	24: TEK
		26: SCA1	25: TPM3
		27: SLC2A1	26: TYRP1
		28: SELE	27: TGFBI
		31: SAA1	28: TSHB
		32: SNCA	29: TNNI3
		33: SOD3	30: TIMP3
		34: SCN1B	31: TECTA
		35: SLC6A4	32: TAP1
		36: SRK	33: TCF14
		37: SLC5A1	36: TH
		39: SLC10A2	37: TSHR
			38: THRB
			39: TAP2
			40: TGFBR2

U	V	W	X
1: UMPS	1: VWF	1: WT1	1: XPA
2: UGB	2: VDR	2: WFS1	2: XDH
3: USH2A	3: VMD2	3: WRN	3: XPC
4: UFD1L	4: VHL	4: WAS	6: XK
5: ugt1d			8: XIST
6: UROD			9: XRCC9
7: UBE3A			
8: UCP3			
9: UROS			
10: UGT1			
Y	Z		
	1: ZIC2		
	2: ZIC3		

EXAMPLE 5a
POLYMORPHIC VARIATION

For each gene, sequence data concerning the existence of polymorphic variation can be located. For example, below are the details of the polymorphic variations of six genes, representative of major gene product/protein categories on the core list.

Category 1 - Enzymes

α-glucosidase

Mutation type	Total number of mutations
Nucleotide substitutions (missense / nonsense)	20
Nucleotide substitutions (splicing)	4
Nucleotide substitutions (regulatory)	0
Small deletions	7
Small insertions	0
Small indels	0
Gross deletions	1
Gross insertions & duplications	0
Complex rearrangements (including inversions)	1
Repeat variations	0
TOTAL	33

Accession Number	Codon	Nucleotide	Amino acid	Phenotype
CM970540	40	cCGA-TGA	Arg-Term	Glycogen storage disease 2
CM950491	299	CTG-CGG	Leu-Arg	Glycogen storage disease 2
CM980577	309	cGGG-AGG	Gly-Arg	Glycogen storage disease 2
CM910167	318	ATG-ACG	Met-Thr	Glycogen storage disease 2
CM900102	402	aTGG-CGG	Trp-Arg	Glycogen storage disease 2
CM940798	519	cATG-GTG	Met-Val	Glycogen storage disease 2
CM910168	521	cGAG-AAG	Glu-Lys	Glycogen storage disease 2
CM940799	545	CCT-CTT	Pro-Leu	Glycogen storage disease 2

CM980578	566	cTCC-CCC	Ser-Pro	Glycogen storage disease 2
CM930287	643	cGGG-AGG	Gly-Arg	Glycogen storage disease 2
CM940800	645	GACg-GAA	Asp-Glu	Glycogen storage disease 2
CM980579	645	cGAC-AAC	Asp-Asn	Glycogen storage disease 2
CM950492	645	cGAC-CAC	Asp-His	Glycogen storage disease 2
CM940801	647	TGCg-TGG	Cys-Trp	Glycogen storage disease 2
CM980580	648	cGGC-AGC	Gly-Ser	Glycogen storage disease 2
CM980581	672	CGG-CAG	Arg-Gln	Glycogen storage disease 2
CM980582	672	gCGG-TGG	Arg-Trp	Glycogen storage disease 2
CM930288	725	cCGG-TGG	Arg-Trp	Glycogen storage disease 2
CM980583	768	CCC-CGC	Pro-Arg	Glycogen storage disease 2
CM930289	854	cCGA-TGA	Arg-Term	Glycogen storage disease 2

Accession Number	IVS	Donor/ Acceptor	Relative location	Substitution	Phenotype
CS941486	1	as	-13	T-G	Glycogen storage disease 2
CS971665	6	as	-22	T-G	Glycogen storage disease 2
CS941487	10	ds	+1	G-C	Glycogen storage disease 2
CS971666	16	ds	+2	T-C	Glycogen storage disease 2

Accession Number	Location/ codon	Deletion	Phenotype
CD981927	126	GCAGCCC^TGGtgCTTCTTCCCA	Glycogen storage disease 2
CD972136	160	CACCTTC^TTCccCAAGGACATC	Glycogen storage disease 2
CD941678	174	TGATG^GAGACtGAGAACCGCC	Glycogen storage disease 2
CD961963	470	CATCACC^AACgagaCCGGCCAGCC	Glycogen storage disease 2
CD941679	485	CGGGTCC^ACTgccttccccgactTCACCAACCC	Glycogen storage disease 2
CD981928	674	CGGAAC^CACAacaGCCTGCTCAG	Glycogen storage disease 2
CD951684	902	GCAGCTG^CAGaagGTGACTGTCC	Glycogen storage disease 2

Description	Phenotype
536 bp I17E18-332 to E18I19+39 (mutation described at genomic DNA level)	Glycogen storage disease 2

Description	Phenotype
Ins C nt. 2741, ins G nt. 2743	Glycogen storage disease 2

Category 2 - Transport and Storage

Albumin

Mutation type	Total number of mutations
Nucleotide substitutions (missense / nonsense)	21
Nucleotide substitutions (splicing)	2
Nucleotide substitutions (regulatory)	0
Small deletions	2
Small insertions	1
Small indels	0
Gross deletions	0
Gross insertions & duplications	0
Complex rearrangements (including inversions)	0
Repeat variations	0
TOTAL	26

Accession Number	Codon	Nucleotide	Amino acid	Phenotype
CM910024	1	GAT-GTT	Asp-Val	Albumin variant

CM940018	3	aCAC-TAC	His-Tyr	Albumin variant
CM910025	-1	CGA-CAA	Arg-Gln	Albumin variant
CM910026	-2	CGT-CAT	Arg-His	Albumin variant
CM900011	-2	tCGT-TGT	Arg-Cys	Albumin variant
CM940019	32	tCAG-TAG	Gln-Term	Analbuminaemia
CM940020	114	cCGA-TGA	Arg-Term	Analbuminaemia
CM910027	128	CAT-CGT	His-Arg	Albumin variant
CM940021	214	TGGg-TGA	Trp-Term	Analbuminaemia
CM920015	218	CGC-CAC	Arg-His	Albumin variant
CM970070	218	CGC-CCC	Arg-Pro	Dysalbuminaemic hyperthyroxinaemia, familial
CM940022	225	cAAA-CAA	Lys-Gln	Albumin variant
CM940023	276	AAGg-AAC	Lys-Asn	Albumin variant
CM940024	313	AAGg-AAT	Lys-Asn	Albumin variant
CM910028	365	GAT-GTT	Asp-Val	Albumin variant
CM910029	372	cAAA-GAA	Lys-Glu	Albumin variant
CM900012	501	aGAG-AAG	Glu-Lys	Albumin variant
CM930016	505	tGAA-AAA	Glu-Lys	Albumin variant
CM940025	563	cGAT-AAT	Asp-Asn	Albumin variant
CM910030	570	cGAG-AAG	Glu-Lys	Albumin variant
CM940026	573	tAAA-GAA	Lys-Glu	Albumin variant

Accession Number	Location/ codon	Deletion	Phenotype
CD941562	566	TAAGGAG^ACCtGCTTTGCCGA	Albumin variant
CD910474	579	TGCTGCA^AGTcAAGCTGCCTT	Analbuminaemia

Accession Number	Nucleotide	Codon	Insertion	Phenotype
CI941818	9156	267	A	Analbuminaemia

Category 3 - Structural Proteins

Collagen IV alpha 3

Mutation type	Total number of mutations
Nucleotide substitutions (missense / nonsense)	2
Nucleotide substitutions (splicing)	1
Nucleotide substitutions (regulatory)	0
Small deletions	2
Small insertions	0
Small indels	0
Gross deletions	0
Gross insertions & duplications	0
Complex rearrangements (including inversions)	0
Repeat variations	0
TOTAL	5

Accession Number	Codon	Nucleotide	Amino acid	Phenotype
CM940306	1481	aCGA-TGA	Arg-Term	Alport syndrome
CM940307	1524	TCA-TGA	Ser-Term	Alport syndrome

Accession Number	IVS	Donor/ Acceptor	Relative location	Substitution	Phenotype
CS951356	5	as	-320	G-T	Alport syndrome

Accession Number	Location/ codon	Deletion	Phenotype
CD951631	1448	TTTGTC^TTCAcccgacaCAGTCAAACC	Alport syndrome
CD941648	1471	AGTGGGT^TTTctttttCTTTTTGTAC	Alport syndrome

Category 4 - Immune Protection and inflammation

Interleukin 4 receptor

Mutation type	Total number of mutations
Nucleotide substitutions (missense / nonsense)	1
Nucleotide substitutions (splicing)	0
Nucleotide substitutions (regulatory)	0
Small deletions	0
Small insertions	0
Small indels	0
Gross deletions	0
Gross insertions & duplications	0
Complex rearrangements (including inversions)	0
Repeat variations	0
TOTAL	1

Accession Number	Codon	Nucleotide	Amino acid	Phenotype
CM970744	576	CAG-CGG	Gln-Arg	Atopy, association with

Category 5 – Generation and Transmission of Nervous Impulses

Prion protein

Mutation type	Total number of mutations
Nucleotide substitutions (missense / nonsense)	14
Nucleotide substitutions (splicing)	0
Nucleotide substitutions (regulatory)	0
Small deletions	0
Small insertions	0
Small indels	0
Gross deletions	0
Gross insertions & duplications	0
Complex rearrangements (including inversions)	0
Repeat variations	0
TOTAL	14

Accession Number	Codon	Nucleotide	Amino acid	Phenotype
CM890102	102	CCG-CTG	Pro-Leu	Gerstmann-Straeussler syndrome
CM930595	105	CCA-CTA	Pro-Leu	Gerstmann-Straeussler syndrome
CM890103	117	GCA-GTA	Ala-Val	Gerstmann-Straeussler syndrome
CM890104	129	cATG-GTG	Met-Val	Gerstmann-Straeussler syndrome
CM971202	171	AAC-AGC	Asn-Ser	Schizophrenia
CM910305	178	cGAC-AAC	Asp-Asn	Creutzfeld-Jakob syndrome
CM930596	180	cGTC-ATC	Val-Ile	Creutzfeld-Jakob syndrome

CM971203	183	cACA-GCA	Thr-Ala	Spongiform encephalopathy, familial
CM920588	198	TTC-TCC	Phe-Ser	Gerstmann-Straeussler syndrome
CM890105	200	cGAG-AAG	Glu-Lys	Creutzfeld-Jakob syndrome
CM961133	208	CGC-CAC	Arg-His	Creutzfeld-Jakob syndrome
CM930597	210	gGTT-ATT	Val-Ile	Creutzfeld-Jakob syndrome
CM920589	217	CAG-CGG	Gln-Arg	Gerstmann-Straeussler syndrome
CM930598	232	ATG-AGG	Met-Arg	Creutzfeld-Jakob syndrome

Category 6 - Growth and Differentiation

Vitamin D receptor

Mutation type	Total number of mutations
Nucleotide substitutions (missense / nonsense)	10
Nucleotide substitutions (splicing)	1
Nucleotide substitutions (regulatory)	0
Small deletions	0
Small insertions	0
Small indels	0
Gross deletions	0
Gross insertions & duplications	0
Complex rearrangements (including inversions)	0
Repeat variations	0
TOTAL	11

Accession Number	Codon	Nucleotide	Amino acid	Phenotype
CM971505	30	cCGA-TGA	Arg-Term	Rickets, vitamin D resistant
CM880062	33	GGC-GAC	Gly-Asp	Rickets, vitamin D resistant
CM961380	46	GGC-GAC	Gly-Asp	Rickets, vitamin D resistant
CM910389	50	CGA-CAA	Arg-Gln	Rickets, vitamin D resistant
CM880063	73	CGA-CAA	Arg-Gln	Rickets, vitamin D resistant
CM900227	80	CGG-CAG	Arg-Gln	Rickets, vitamin D resistant
CM930718	152	cCAG-TAG	Gln-Term	Rickets, vitamin D resistant
CM930719	274	CGC-CTC	Arg-Leu	Rickets, vitamin D resistant
CM890115	295	TACc-TAA	Tyr-Term	Rickets, vitamin D resistant
CM971506	305	CACa-CAG	His-Gln	Rickets, vitamin D resistant

Accession Number	IVS	Donor/ Acceptor	Relative location	Substitution	Phenotype
CS961654	4	ds	+5	G-C	Rickets, vitamin D resistant

The identification of the core group of genes considered to have an important effect on the physiological and pathophysiological processes of disease enables attention to be focussed on ascertaining, identifying and cataloguing the genetic vatriation within the core group of genes utilising tried and tested technologies and techniques.

EXAMPLE 6
IDENTIFYING AND DETECTING POLYMORPHIC VARIATION IN THE CORE LIST OF GENES

The human genome is known to be highly variable in different individuals. Variation exists in approximately one nucleic acid residue in every 300. Although a single

nucleic acid change (single nucleotide polymorphism, SNP e.g. Schafer and Hawkins 1997, Nickerson et al 1998, Rieder et al 1998, SNP Consortium 1999) is the commonest form of genetic variation, other more complex forms also occur for example:

Type of variation	Example
Deletion	intronic deletion in the angiotensin converting enzyme gene
Insertion	144bp insertion in the prion gene
Repeats	Huntingtin gene in Huntington's chorea

These more complex forms of genetic variations account for more than 40% of the genetic changes associated with human disease.

Variations in human gene sequences, which are present in more than 1% of the population, are known as polymorphisms. These changes in genetic sequence can be detected by a variety of methods, which allow the direct sequencing and correct alignment of nucleotides (e.g. the Sanger method). However, this method is prone to error and multiple runs are required to ensure accuracy. More recently (Schafer and Hawkins 1997, Gilles et al 1999) many other techniques have been developed to, accurately and sensitively, identify the presence of polymorphic variation based on:

- restriction fragment length polymorphisms using Southern blots
- allele specific extensions of a detection primer using high fidelity enzymes
- scanning for single strand conformational polymorphisms
- gel mobility detection of heteroduplexs
- detection of denaturing gradient differences using gel electrophoresis
- ribonuclease cleavage of RNA:RNA or RNA:DNA heteroduplexes
- chemical cleavage of heteroduplex mismatches
- gel based detection of resolvase cleavage using T4 endonuclease
- radioactive labelling and multi-photon detection
- detection of altered banding patterns on gels using cleavage fragment length polymorphisms
- recognition of heteroduplex mismatches using E. Coli mismatch repair enzymes
- DNA variation detection using denaturing high performance liquid chromatography
- matrix assisted laser desorption/ionisation time of flight mass spectrometry

- electronic array of DNA probes on silicon microchips

Therefore, given an identified gene sequence, the technology to identify polymorphic variation is well established and is generally applicable to any section of the human genome. (Nickerson et al 1998, Wang et al 1998, Rieder et al 1999).

In addition computational approaches can also be used to search for and assess polymorphic variation in existing gene sequence databases (as confirmed by Buetow et al 1999).

Thus the methods of generating the nucleotide sequence required for the design of an array or chip is well known to those skilled in the art.

However, for the purposes of an array design it would be useful to establish the frequency of a given polymorphism in the general population and thus derive a way of assessing its likely clinical importance. Polymorphisms are defined as being a genetic variation present in more than 1% of the population. In order to determine the frequency of a polymorphism in a given population a number of individual DNA samples will need to be investigated. The table below provides the number of DNA samples, which will need to be examined in order to determine the frequency of polymorphisms at a particular threshold of statistical certainty.

NUMBER OF DNA SAMPLES REQUIRED TO DETECT POLYMORPHISMS

Minimum Allele Frequency	**Appears Once**	**Appears Twice**	**Statistical Certainty**
> 1%	58	97	90%
	75	119	95%
	115	166	99%
> 5%	12	19	90%
	15	24	95%
	23	33	99%
> 10%	6	10	90%
	8	12	95%
	11	16	99%

E.g. if a particular variant appears twice in 166 DNA samples, we can be 99% sure that the variant allele is present in >1% of the population.

The technologies and methodologies required for the identification and tabulation of polymorphic variation are of considerable value in the identification of genetic variation, which will be informative in the practice of medicine.

This invention provides a means of fusing the genomic and pharmacological profiles together with their clinical associations in such a way as to enhance and enable the provision of individually tailored therapeutic packages for enhanced healthcare management.

In addition, the use of such devices and the tabulating of genomic variations that lead to or predispose to disease, will lead to revolutionary insights into the pathophysiology of diseases. These may well lead to the classical definitions of disease states being sub-divided or re-organised into specific genomic configurations,

creating the potential for new therapeutic approaches (as indicated in Drews and Ryser 1997).

The actual demonstration of associations between disease, outcomes, adverse events or specific symptom clusters will emerge as the result of clinical trials and investigations using accepted approaches and methods.

EXAMPLE 7 - ANALYSIS OF DATABASE TO ASCERTAIN GENOTYPE/PHENOTYPE RELATIONSHIPS

The generation of genetic profiling data and its analysis alongside clinical information derived from patients presents considerable challenges for data handling and analysis. The volume of information, number of information categories and the variable nature of the information (e.g. dimensional or categorical) ensure that the operation of a database combining genetic and clinical information to generate a prognostic outcome is a complex task.

However, the complexity can be dealt with using existing analytical approaches. Association analysis between genetic polymorphisms can be dealt with by using standard statistical techniques (analysis of variance, meta-analysis etc) with appropriate corrections for multiple testing. The thresholds for statistical significance will be derived from scientific convention (e.g. significance at the 5% level following Bonnferoni correction). The data concerning genotype/phenotype relationships between the core group of genes and clinical signs and symptoms and therapeutic interventions will form a central component of the database.

The creation of a database containing and elaborating on such genotype/phenotype relationships will become an important tool for the practice of molecular medicine and the development of healthcare management. In order to derive benefit from such a database it must be capable (following interrogation using a patients profile of genetic variation derived from the core group of genes) of analysing the profile and providing a meaningful output to the healthcare professional which will provide guidance on the prognosis, healthcare management and therapeutic interventions appropriate to the patient.

The generation of such an output can be achieved using machine learning algorithms. The genetic algorithm (Goldberg 1989, Fogarty and Ireson 1994) has been shown to provide a general process for achieving good results for search in large noisy domains. Starting from a population of randomly generated points in a search space, and given an evaluation of each of those points, the genetic algorithm is designed to converge the population to an optimum point in the search space. Processes of data selection, crossover, mutation and replacement of old members of the dataset achieve this with new members of more value. The effective use of the genetic algorithm process is a representation of the search space, which is responsive to the heuristics, embodied in the genetic operators.

The user must also supply an evaluation function identifying the degree to which the point in space approaches an optimum ('weighting') such that the selection operator for propagation through the dataset can choose them.
The genetic algorithm can be used to find predictively meaningful categories that is:

- intervals of continuous attribute values
- sets of nominal attribute values
- combinations of attributes

Together these attributes can create a simple Bayesian classifier for aspects of healthcare management.

Additional techniques (e.g. Bahadur-Lazarsfeld expansion) enable second order approximation of dependencies between predictive attributes. This allows the full complexity of the individual's genetic variation profile and the specifics of their clinical, psychological and social state to be assessed in order to produce an output concerning their prognosis, healthcare management and the possibilities for therapeutic intervention.

Assembly of such data will allow the merging of accepted treatment algorithms with the polymorphic variation underlying specific aspects of genomic functionality. This will produce new algorithms that will provide a prognostic indication for individual patients and, coupled with the expertise of their responsible clinician, allow the appropriate healthcare decisions to be made in a pro-active way.

The identification of genetic variation in the core list of genes and its application to healthcare management will have significant beneficial effects on the way in which clinicians will be able to formulate plans for healthcare management.

This will be seen in at least two ways. The first by enabling the targeting of resources at appropriate individuals (see Example 8) and the second by enabling an objective risk assessment of the optimum configuration for different types of therapeutic intervention (e.g drugs, surgery, radiotherapy, occupational therapy) and the identification of those patients at significant risk of suffering adverse events from therapeutic intervention (see Example 9).

EXAMPLE 8 - CLINICAL MANAGEMENT OF FAMILIAL ADEMATOUS POLYPOSIS

Familial adenomatous polyposis (FAP) is an autosomal dominant disorder which typically presents with colorectal cancer (CRC) in early adult life secondary to extensive adenomatous polyps of the colon. Polyps also develop in the upper gastrointestinal tract and malignancies may occur in other sites including the brain and the thyroid. Helpful diagnostic features include pigmented retinal lesions known as congenital hypertrophy of the retinal pigment, jaw cysts, sebaceous cysts, and osteomata. The APC gene at 5q21 is mutant in FAP.

CLINICAL FEATURES

Familial adenomatous polyposis (FAP) is characterized by adenomatous polyps of the colon and rectum; in extreme cases the bowel is carpeted with a myriad of polyps. This is an aggressive premalignant disease with one or more polyps progressing through dysplasia to malignancy in untreated gene carriers with a median age at diagnosis of 40 years. Carcinoma may arise at any age from late childhood through

the seventh decade. The presenting features are usually those of malignancy, such as weight loss and inanition, bowel obstruction, or bloody diarrhea. Cases of new mutation still present in these ways but in areas with well organized registers most other gene carriers are detected by bowel examination while still asymptomatic. Occasionally, the extracolonic features of the condition lead to presentation.

Petersen et al. (1993) demonstrated the feasibility of presymptomatic direct detection of APC mutations in each of 4 families. No change in the conventional FAP colon screening regimen was recommended for children found to have a mutation. In contrast, when direct tests indicated that an individual did not have the mutation, they recommended that screening be decreased. Three of the mutations were nonsense mutations and one was a frameshift mutation due to insertion of 1 nucleotide. In an evaluation of molecular genetic diagnosis in the management of familial polyposis, Maher et al. (1993) concluded that intragenic and closely linked DNA markers are informative in most families and that, in addition to the clinical benefits of presymptomatic diagnosis, the reduction in screening for low-risk relatives means that molecular genetic diagnosis is a cost-effective procedure.

Davies et al. (1995) found that families with mutations 3-prime of codon 1444 had significantly more lesions on dental panoramic radiographs (P less than 0.001) and appeared to have a higher incidence of desmoid tumors than did families with mutations at the 5-prime end. All 7 families except one with mutations 5-prime of exon 9 did not express CHRPE. All of 38 individuals from 16 families with mutations between exon 9 and codon 1444 expressed CHRPE. The 11 individuals from 4 families with mutations 3-prime of codon 1444 did not express CHRPE. These results suggested that the severity of some of the features of Gardner syndrome may correlate with genotype in FAP.

Since an alteration of the APC gene occurs early in most colorectal tumors, detection of APC mutations in fecal tumor DNA could be a powerful tool for the diagnosis of noninvasive cancer. Deuter and Muller (1998) described a highly sensitive and nonradioactive heteroduplex-PCR method (HD-PCR) for detecting APC mutations in stool DNA.

Petersen et al. (1989) demonstrated how one could use linkage information to modify the standard recommendations for follow-up. For example, in the family of an affected 36-year-old man with a positive family history of APC, there were 4 asymptomatic children under the age of 10 years. Before linkage analysis, all children had a 50% risk. Screening protocols would call for annual sigmoidoscopy in all beginning at age 12 years. With the linkage information, one could state to the family with 98% confidence that 3 of the children did not inherit the gene and that 1 child did. That child could be screened annually; the others would have screening every 3 years beginning at ages 12 or 13 and continuing until age 35.

EXAMPLE 9 - GENETIC VARIATION IN DRUG TARGETS AND DRUG METABOLIZING ENZYMES

Therapeutic intervention by the use of drugs is a common mode of clinical treatment. However, this is not without difficulty (Weatherall, Leadingham and Warell 1996) and even hazard (Lazarou et al 1998). Drugs interact with the body in many different

ways to produce their effect. Some drugs act as false substrates of inhibitors for transport systems (e.g. calcium channels) or enzymes (acetylcholinesterase). Most drugs however, produce their effects by acting on receptors, usually located in the cell membrane, which normally respond to endogenous chemicals in the body (Weatherall, Leadingham and Warrell 1996). Drugs that activate receptors and produce a response are called agonists (e.g cholinomimetics). Antagonists combine with receptors but do not activate them, thus reduceing the probability of the transmitter substance combining with the receptor and so blocking receptor activation. The ability of the drug to interact with the receptor depends on the specificity of the drug for the receptor or 'target' (Brody, Larner and Minneman 1998).

In addition to the main categories of agonist and antagonist, drugs also have mechanisms of action whereupon they interact with specific types of molecules - targets' - that include:

- blockade of uptake or transport sites (e.g selective serotonin reuptake inhibitors)
- enzyme inhibition (e.g. angiotensin convertying enzyme inhibitors, acetylcholinesterase inhibitors)
- blockade of ion channels (calcium channel antagonists, anaesthetics)

However, many drugs are known to vary in their efficacy and side effects from patient to patient. This variation in drug response will be associated with the polymorphic variation in the drug target.

CNS MARKETED DRUGS

Drug	Drug Target	Polymorphic?
Tricyclic antidepressants (TCA)	Neurotransmitter (NA/5-HT) re-uptake proteins (NET & SERT)	✓
SSRIs	Selective serotonin transport re-uptake protein (SERT)	✓
MAOIs	Monoamine oxidase A & B	✓
Benzodiazepines (GABA facilitators)/GABA antagonists. Barbiturates.	GABA receptors	✓
Beta-blockers	Noradrenaline (beta-adrenergic) receptors	✓
Atypical antidepressants	Alpha-adrenoceptors	✓
Beta-adrenoceptors antagonists	Beta-adrenoceptors	
Dopamine blockers/ boosters	Dopamine receptors	✓
Dopamine blockers/ boosters/depleters	Dopamine transporter (DAT1)	✓
Anticholinergics (muscarinic antagonists)	Muscarinic receptors	✓
Anticholinergics (nicotinic antagonists)	Nicotinic receptors	✓
Anticholinesterases	Acetylcholinesterase (ACHE)	✓
COMT inhibitor	Catechol-O-methyltransferase (COMT)	✓
Sodium channel blocker	Sodium channel	✓

Opioid analgesics & antagonists	Opioid receptors (OPRM1; OPRK1; OPRD1)	✓
Antipsychotics/neuroleptics (5-HT/D2 antagonists)	5-HT/D2 receptors	✓
Antiinflammatory drugs	Cyclooxygenase (COX1, COX2)	✓
Antihistamines	Histamine receptors	✓

CARDIOVASCULAR MARKETED DRUGS

Drug	Drug Target	Polymorphic?
ACE inhibitors	Angiotensin converting enzyme (ACE)	✓
HMG CoA reductase inhibitors, e.g simvastatin	HMG CoA reductase	✓
Angiotensin II antagonists	Angiotensinogen	✓
Calcium channel blocker	Calcium channel	✓
Thromboxane A2 synthase inhibitor	Thromboxane A2 synthase	✓
A2 receptor antagonist	Thromboxane A2 receptor	✓
Potassium channel blocker	Potassium channel	✓
Na-H ion exchange (NHE) inhibitor	Na-H ion exchanger (NHE)	✓
bile acid transport inhibitor	SLC10A1 (sodium/bile acid cotransporter)	✓
bile acid transport inhibitor	SLC10A2 (sodium/bile acid cotransporter)	✓
platelet aggregation inhibitor	Von Willebrand factor	✓
ACAT inhibitor	Acetoacetyl-CoA-thiolase (ACAT)	✓
Endothelin antagonist	Endothelin (EDN3)	✓

GASTROINTESTINAL (Peptic ulcer) MARKETED DRUGS

Drug	Drug Target	Polymorphic?
Proton pump inhibitor (e.g omeprazole).	H+/K+ adenosine triphosphatase (ATPase) enzyme system ('proton pump')	✓
H2 antagonists (e.g.cimetidine)	Histamine H2-receptor	✓
Muscarinic antagonists (e.g.pirenepine)	Muscarinic m1 & m3 receptors	✓
Prostaglandins (inhibit cAMP)	Adenylate cyclase, histamine-induced activity	✓

Another problem the medical practitioner faces, is that certain patients may be particularly susceptible to drug addiction. Examples of drugs with known addictive properties are Amphetamines, Temazepam and Phenobarbitone, although having approved medicinal use e.g. phenobarbitone for epilepsy, they may cause problems of dependency and misuse in individuals. Knowledge of such an individual's susceptibility before prescribing certain drugs would be an advantage to the medical practitioner.

Any drug may produce unwanted or unexpected adverse events, these can range from trivial (slight nausea) to fatal (aplastic anaemia). One of the main reasons for adverse

events following drug intake is the drug binding to a non specific or non target receptors in the body (Brody, Larner and Minneman 1998). Another reason is the interaction of the drug with other drugs given to the patient. This is a particular problem in the elderly who frequently suffer from multiple illnesses requiring many different classes of drugs and providing a real potential for drug interactions (Weatherall, Leadingham and Warrell 1996). The drug may also produce adverse events over time as the drug is absorbed, distributed, metabolised and excreted e.g. products of metabolising the drug may be reactive themselves and be toxic to the body. Being able to predict the likelihood of a particular individual suffering from an adverse event and the severity of that event would be an important tool for the practitioner. Many of the important components of the biological pathways involved in drug metabolism are coded by genes containing polymorphic variation.

METABOLISING ENZYMES

Drug	Drug-metabolising enzyme	Polymorphic?
Most	Cytochrome P450 enzyme, CYP2C19	✓
Most	Cytochrome P450 enzyme, CYP2D6	✓
Most	UDP-glucuronosyltransferase	✓
Most	N-acetyltransferase (NAT1)	✓
Most	Methyltransferase	✓
Most	Sulphotransferase	✓
Most	NADPH-cytochrome p450 reductase	✓

The inventory of drugs and preparations both registered and in development which can be matched to drug targets exhibiting genetic polymorphisms can be found in standard works of reference, in particular the British National Formulary, 1998, the Dental Practioners' Formulary, 1998, Martindale, 1998, Herbal medicines, 1998. Drugs available in the United States can be found in U.S. Pharmacopeia, 1998, and drugs available in Japan can be found in Iryoyaku Nihon Iyakuhinshu, 1998, Ippanyaku Nihon Iyakuhinshu, 1998 and Hokenyaku Jiten, 1998. Drugs available in other countries can be found in the appropriate National Formularies. A list of drugs currently under development worldwide can be found in current journals and text (Pipeline pulse, 1999, Scrip, 1998, IDrugs, 1998, Current Opinion in Drug Discovery and Development, 1998).

The use of the Genostic approach described above would be of considerable utility in determining the likelihood and magnitude of therapeutic response to drugs in the inventories described above. Such difficulties can arise from adverse events, variations in metabolism and drug-drug interactions in situations where several diseases, requiring treatment, exist in a given patient. The potential for adverse events or deleterious outcomes could be ascertained in individuals, patients or populations in relation to all of the drugs referred to above. These factors are of considerable importance in enabling the selection and monitoring of therapeutic interventions and effective healthcare management.

CORE GENES FOR DESIGN AND MANUFACTURE OF 'GENOSTICS'

We have elaborated on the value and utility to be derived from the gathering together of the genes which form the core gene list for the Genostic system.

These genes are elaborated below:

KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME
T TRANSPORT & STORAGE
S STRUCTURAL
I IMMUNITY
N NERVOUS TRANSMISSION
G GROWTH & DIFFERENTIATION

CORE GENE LIST	HUGO GENE SYMBOL	PROTEIN FUNCTION
11beta hydroxysteroid dehydrogenase 2	HSD11B2	E
17beta hydroxysteroid dehydrogenase 1	HSD17B1	E
17beta hydroxysteroid dehydrogenase 3	HSD17B3	E
17beta hydroxysteroid dehydrogenase 4	HSD17B4	E
17beta hydroxysteroid oxidoreductase		E
18-hydroxysteroid oxidoreductase		E
2,3-bisphosphoglycerate mutase	BPGM	E
2,4-dienoyl CoA reductase	DECR	E
3 beta hydroxysteroid dehydrogenase 2	HSD3B2	E
3-oxoacid CoA transferase	OXCT	E
4-hydroxyphenylpyruvate dioxygenase	HPD	E
5,10-methylenetetrahydrofolate reductase (NADPH)	MTHFR	E
5-adenosyl homocysteine hydrolase		E
6-phosphofructo-2-kinase	PFKFB1	E
6-pyruvoyltetrahydropterin synthase	PTS	E
Acetoacetyl 1-CoA-thiolase	ACAT1	E
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetyl CoA acyltransferase	ACAA	E
Acetyl CoA carboxylase	ACC	E
Acetyl CoA carboxylase alpha	ACACA	E
Acetyl CoA synthase		E
Acetylcholinesterase	ACHE	E
Acid phosphatase 2, lysosomal	ACP2	E
Aconitase		E
Acyl CoA dehydrogenase, long chain	ACADL	E
Acyl CoA dehydrogenase, medium chain	ACADM	E
Acyl CoA dehydrogenase, short chain	ACADS	E
Acyl CoA dehydrogenase, very long chain	ACADVL	E
Acyl CoA synthetase, long chain, 1	LACS1	E
Acyl CoA synthetase, long chain, 2	LACS2	E

Acyl CoA synthetase, long chain, 4	ACS4	E
Acyl malonyl condensing enzyme		E
Acyl-CoA thioesterase		E
ADAM (A disintegrin and metalloproteinase) 1	ADAM1	E
ADAM (A disintegrin and metalloproteinase) 10	ADAM10	E
ADAM (A disintegrin and metalloproteinase) 11	ADAM11	E
ADAM (A disintegrin and metalloproteinase) 12	ADAM12	E
ADAM (A disintegrin and metalloproteinase) 13	ADAM13	E
ADAM (A disintegrin and metalloproteinase) 14	ADAM14	E
ADAM (A disintegrin and metalloproteinase) 15	ADAM15	E
ADAM (A disintegrin and metalloproteinase) 16	ADAM16	E
ADAM (A disintegrin and metalloproteinase) 17	ADAM17	E
ADAM (A disintegrin and metalloproteinase) 18	ADAM18	E
ADAM (A disintegrin and metalloproteinase) 19	ADAM19	E
ADAM (A disintegrin and metalloproteinase) 2	ADAM2	E
ADAM (A disintegrin and metalloproteinase) 3A	ADAM3A	E
ADAM (A disintegrin and metalloproteinase) 3B	ADAM3B	E
ADAM (A disintegrin and metalloproteinase) 4	ADAM4	E
ADAM (A disintegrin and metalloproteinase) 5	ADAM5	E
ADAM (A disintegrin and metalloproteinase) 6	ADAM6	E
ADAM (A disintegrin and metalloproteinase) 7	ADAM7	E
ADAM (A disintegrin and metalloproteinase) 8	ADAM8	E
ADAM (A disintegrin and metalloproteinase) 9	ADAM9	E
Adenosine deaminase	ADA	E
Adenosine monophosphate deaminase	AMPD	E
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adenylate kinase	AK1	E
Adenylate transferase		E
Adenylosuccinate lyase	ADSL	E
ADP-ribosyltransferase	ADPRT	E
Adrenoleukodystrophy gene	ALD	E
Alanine-glyoxylate aminotransferase	AGXT	E
Alcohol dehydrogenase 1	ADH1	E
Alcohol dehydrogenase 2	ADH2	E
Alcohol dehydrogenase 3	ADH3	E
Alcohol dehydrogenase 4	ADH4	E
Alcohol dehydrogenase 5	ADH5	E
Alcohol dehydrogenase 6	ADH6	E
Alcohol dehydrogenase 7	ADH7	E
Aldehyde dehydrogenase 1	ALDH1	E
Aldehyde dehydrogenase 10	ALDH10	E
Aldehyde dehydrogenase 2	ALDH2	E

Aldehyde dehydrogenase 5	ALDH5	E
Aldehyde dehydrogenase 6	ALDH6	E
Aldehyde dehydrogenase 7	ALDH7	E
Aldolase A	ALDOA	E
Aldolase B	ALDOB	E
Aldolase C	ALDOC	E
Alkylglycerone phosphate synthase	AGPS	E
alpha1-antichymotrypsin	AACT	E
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
alpha-amino adipic semialdehyde synthase		E
alpha-amylase		E
alpha-dextrinase		E
alpha-Galactosidase A	GLA	E
Alpha-galactosidase B, GALB	NAGA	E
alpha-glucosidase, neutral C	GANC	E
alpha-glucosidase, neutral AB	GANAB	E
Peptidylglycine alpha-amidating monooxygenase	PAM	E
alpha-ketoglutarate dehydrogenase		E
alpha-L-Iduronidase	IDUA	E
Aminomethyltransferase	AMT	E
Aminopeptidase P	XPNPEP2	E
Amylo-1,6-glucosidase	AGL	E
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensinogen	AGT	E
Antithrombin III	AT3	E
Apurinic endonuclease	APE	E
Arginase	ARG1	E
Arginosuccinate lyase	ASL	E
Arginosuccinate synthetase	ASS	E
Arylsulfatase A	ARSA	E
Arylsulfatase B	ARSB	E
Arylsulfatase C	ARSC1	E
Arylsulfatase D	ARSD	E
Arylsulfatase E	ARSE	E
Arylsulfatase F	ARSF	E
Asparagine synthetase	AS	E
Aspartate transcarbamoylase		E
Aspartoacylase	ASPA	E
Aspartylglucosaminidase	AGA	E
ATP cobalamin adenoxyltransferase		E
ATP sulphurylase	atpsk2	E
ATP/ADP translocase		E
beta-galactosidase	GLB1	E
beta-glucosidase, neutral		E
beta-Glucuronidase	GUSB	E
beta-ketoacyl reductase		E
beta-N-acetylhexosaminidase, A		E
beta-N-acetylhexosaminidase, B		E
Bile acid coenzyme A: amino acid N-	BAAT	E

acyltransferase		
Bile salt-stimulated lipase	CEL	E
Bilirubin UDP-glucuronosyltransferase		E
Biotinidase	BTD	E
Bleomycin hydrolase	BLMH	E
Branched chain aminotransferase 1, cytosolic	BCAT1	E
Branched chain aminotransferase 2, mitochondrial	BCAT2	E
Branched chain keto acid dehydrogenase E1, alpha polypeptide	BCKDHA	E
Branched chain keto acid dehydrogenase E1, beta polypeptide	BCKDHB	E
Brush border guanylyl cyclase		E
Butyrylcholinesterase	BCHE	E
C1 inhibitor		E
C17-20 desmolase		E
C3 convertase		E
Calpain	CAPN, CAPN3	E
Carbamoylphosphate synthetase 1	CPS1	E
Carbamoylphosphate synthetase 2	CPS2	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carboxylesterase 1	CES1	E
Carboxypeptidase	CPN	E
Carnitine acetyltransferase	CRAT	E
Carnitine acylcarnitine translocase	CACT	E
Carnitine palmitoyltransferase I	CPT1A	E
Carnitine palmitoyltransferase II	CPT2	E
Catechol-O-methyltransferase	COMT	E
Cathepsin B		E
Cathepsin D		E
Cathepsin E		E
Cathepsin G	CTSG	E
Cathepsin H		E
Cathepsin K	CTSK	E
Cathepsin L		E
Cathepsin S		E
Caveolin 3	CAV3	E
Ceruloplasmin precursor	CP	E
Chitotriosidase	chit	E
Cholesterol ester hydroxylase		E
Choline acetyltransferase	CHAT	E
Chymase	CHY1	
Chymotrypsinogen		E
Citrate synthase		E
CoA transferase		E
Coenzyme Q (CoQ)/ubiquinone		E
Collagenic-like tail subunit of asymmetric acetylcholinesterase	COLQ	E

Complex I		E
Complex II		E
Complex III		E
Complex III		E
Complex V	MTATP6	E
Coproporphyrinogen oxidase	CPO	E
Creatine kinase – B and m	CKBE	E
Cu2+ transporting ATPase alpha polypeptide	ATP7A	E
Cu2+ transporting ATPase beta polypeptide	ATP7B	E
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E
CYP2J2	CYP2J2	E

CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cytidine deaminase	CDA	E
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome b-245 alpha	CYBA	E
Cytochrome b-245 beta	CYBB	E
Cytochrome b-5	CYB5	E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
D-beta-hydroxybutyrate dehydrogenase		E
Dehydratase		E
Delta 4-5 alpha-reductase		E
Delta 4-5 oxosteroid isomerase		E
Delta aminolevulinate dehydratase	ALAD	E
Delta aminolevulinate synthase 1	ALAS1	E
Delta aminolevulinate synthase 2	ALAS2	E
Delta(4)-3-oxosteroid 5-beta-reductase		E
Delta-7-dehydrocholesterol reductase	DHCR7	E
Deoxycorticosterone (DOC) receptor		E
Deoxycytidine kinase DCK		E
Deoxyuridine triphosphatase; dUTPase		E
DHEA sulfotransferase	STD	E
Dihydrodiol dehydrogenase 1	DDH1	E
Dihydrofolate reductase	DHFR	E
Dihydrolipoyl dehydrogenase		E
Dihydrolipoyl dehydrogenase 2	PDHA	E
Dihydrolipoyl succinyltransferase	DLST	E
Dihydrolipoyl transacetylase	PDHA	E
Dihydroorotase		E
Dihydropyramidinase	DPYS	E
Dihydroxyacetonephosphate acyltransferase	DHAPAT	E
Dihyropyrimidine dehydrogenase	DPYD	E
DM-Kinase	DMPK	E
DNA directed polymerase, alpha	POLA	E
DNA glycosylases		E
DNA helicases		E
DNA Ligase 1	LIG1	E

DNA methyltransferase	DNMT	E
Methylguanine-DNA methyltransferase	MGMT	E
DNA polymerase 1		E
DNA polymerase 2		E
DNA polymerase 3		E
DNA primase		E
DNA-dependant RNA polymerase		E
DOPA decarboxylase	DDC	E
Dopamine beta hydroxylase	DBH	E
Dysferlin	DYS, DYSF	E
Dystrophia myotonica	DM, DMPK	E
Dystrophia myotonica, atypical	DM2	E
Elastase 1	ELAS1	E
Elastase 2	ELAS2	E
Electron-transferring flavoprotein dehydrogenase	ETFDH	E
Enolase	ENO1	E
Enoyl CoA hydratase		E
Enoyl CoA isomerase		E
Enoyl CoA reductase		E
Enterokinase	PRSS7, ENTK	E
Eosinophil peroxidase	EPX	E
Epilepsy, benign neonatal 4 gene	ICCA	E
Epilepsy, female restricted	EFMR	E
Epilepsy, progressive myoclonic 2 gene	EPM2A	E
Epoxide hydrolase 1, microsomal	EPHX1	E
Excision repair complementation group 1 protein	ERCC1	E
Excision repair complementation group 2 protein	ERCC2	E
Excision repair complementation group 2 protein	ERCC3	E
Excision repair complementation group 4 protein	ERCC4	E
Excision repair complementation group 6 protein	ERCC6	E
FADH dehydrogenase		E
Ferrochelatase	FECH	E
Flavin-containing monooxygenase 1	FMO1	E
Flavin-containing monooxygenase 2	FMO2	E
Flavin-containing monooxygenase 3	FMO3	E
Flavin-containing monooxygenase 4	FMO4	E
Formiminotransferase		E
Fructose-1,6-diphosphatase	FBP1	E
Fucosidase alpha-L-1	FUCA1	E
Fucosidase alpha-L-2		E
Fumarase	FH	E
Fumarylacetoacetase	FAH	E
GABA transaminase	ABAT	E
Gadd45 (growth arrest & DNA-damage-inducible protein)		E
Galactocerebrosidase	GALC	E
Galactokinase	GALK1	E
Galactose 1-phosphate uridyl-transferase	GALT	E
Gastric Intrinsic factor, GIF	GIF	E
Glucokinase	GCK	E
Glucosaminyl (N-acetyl) transferase 2, I-branching	GCNT2	E

enzyme		
Glucose-6-phosphatase	G6PC	E
Glucose-6-phosphatase translocase	G6PT1	E
Glucose-6-phosphate dehydrogenase	G6PD	E
Glucosidase, acid alpha	GAA	E
Glucosidase, acid beta	GBA	E
Glutamate decarboxylase, GAD	GAD1	E
Glutamate dehydrogenase	GLUD1	E
Glutamate-cysteine ligase	GLCLC	E
Glutamine phosphoribosylpyrophosphate amidotransferase/PRPP amidotransferase		E
Glutamine synthase		E
Glutaryl-CoA dehydrogenase	GCDH	E
Glutathione peroxidase, GPX1	GPX1	E
Glutathione peroxidase, GPX2	GPX2	E
Glutathione reductase, GSR	GSR	E
Glutathione S-transferase mu 1, GSTM1	GSTM1	E
Glutathione S-transferase mu 4, GSTM4		E
Glutathione S-transferase theta 1, GSTT1	GSTT1	E
Glutathione S-transferase theta 2, GSTT2		E
Glutathione S-transferase, GSTP1	GSTP1	E
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glutathione synthetase	GSS	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycerophosphate dehydrogenase 2	GPD2	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Glycine dehydrogenase	GLDC	E
Glycogen branching enzyme	GBE1	E
Glycogen phosphorylase	PYGL	E
Glycogen synthase 1 (muscle)	GLYS1	E
Glycogen synthase 2 (liver)	GYS2	E
Glycosyltransferases, ABO blood group	ABO	E
GM2 ganglioside activator protein, GM2A	GM2A	E
Guanidinoacetate N-methyltransferase	GAMT	E
Guanylate cyclase 2D, membrane (retina-specific)	GUCY2D	E
Guanylate cyclase activator 1A (retina)	GUCA1A	E
Guanylate kinase		E
Guanylyl cyclase		E
Haeme regulated inhibitor kinase		E
Heparan sulfamidase		E
Hepatic lipase	LIPC	E
Hepatic nuclear factor-3-beta	HNF3B	E
Hepatic nuclear factor-4-alpha	HNF4A	E
Hexokinase 1	HK1	E
Hexokinase 2	HK2	E
Hexosaminidase A	HEXA,TSD	E
Hexosaminidase B	HEXB	E
Histidase		E

HMG-CoA lyase	HMGCL	E
HMG-CoA reductase	HMGCR	E
HMG-CoA synthase	HMGCS2	E
Holocarboxylase synthetase	HLCS	E
Homogentisate 1,2 dioxygenase	HGD	E
Hormone-sensitive lipase	HSL	E
HSSB, replication protein		E
Hydroxyacyl glutathione hydrolase	HAGH	E
Hypoxanthine-guanine phosphoribosyltransferase, HGPRT	HPRT	E
Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
Ibonucleoside diphosphate reductase		E
Iduronate 2 sulphatase	IDS	E
Inosine monophosphate dehydrogenase, IMPDH		E
Inosine triphosphatase	ITPA	E
Inter-alpha-trypsin inhibitor, IATI		E
Iodothyronine-5'-deiodinase, type 1 and 2		E
IP3 kinase		E
Isocitrate dehydrogenase		E
Isovaleric acid CoA dehydrogenase	IVD	E
Ketohexokinase	KHK	E
ketolase		E
Kynurenine hydroxylase		E
Kynureninease		E
Lactase		E
Lactate dehydrogenase, A	LDHA	E
Lactate dehydrogenase, B	LDHB	E
Lecithin-cholesterol acyltransferase	LCAT	E
Leukotriene A4 synthase	LTA4S	E
Leukotriene B4 synthase	LTB4S	E
Leukotriene C4 synthase	LTC4S	E
Lipoamide dehydrogenase	OGDH	E
Lipoxygenase		E
Lowe oculocerbrorenal syndrome gene	OCRL	E
Lysosomal acid lipase	LIPA	E
Lysyl hydroxylase	PLOD	E
Lysyl oxidase	LOX	E
Malate dehydrogenase, mitochondrial	MDH2	E
Malonyl CoA decarboxylase		E
Malonyl CoA transferase		E
Maltase-glucoamylase		E
Mannosidase, alpha B lysosomal	MANB	E
Mannosidase, beta A lysosomal	MANBA	E
Matrix metalloproteinase 1	MMP1	E
Matrix metalloproteinase 10	MMP10	E
Matrix metalloproteinase 11	MMP11	E
Matrix metalloproteinase 12	MMP12	E
Matrix metalloproteinase 13	MMP13	E
Matrix metalloproteinase 14	MMP14	E

Matrix metalloproteinase 15	MMP15	E
Matrix metalloproteinase 16	MMP16	E
Matrix metalloproteinase 17	MMP17	E
Matrix metalloproteinase 18	MMP18	E
Matrix metalloproteinase 19	MMP19	E
Matrix metalloproteinase 2	MMP2	E
Matrix metalloproteinase 3	MMP3, STMY1	E
Matrix metalloproteinase 4	MMP4	E
Matrix metalloproteinase 5	MMP5	E
Matrix metalloproteinase 6	MMP6	E
Matrix metalloproteinase 7	MMP7	E
Matrix metalloproteinase 8	MMP8	E
Matrix metalloproteinase 9	MMP9	E
MEK kinase, MEKK		E
Methionine adenosyltransferase	MAT1A, MAT2A	E
Methionine synthase	MTR	E
Methionine synthase reductase	MTRR	E
Methylmalonyl-CoA mutase	MUT	E
Mevalonate kinase	MVK	E
Mitochondrial trifunctional protein, alpha subunit	HADHA	E
Mitochondrial trifunctional protein, beta subunit	HADHB	E
Molybdenum cofactor synthesis 1	MOCS1	E
Molybdenum cofactor synthesis 2	MOCS2	E
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Mucolipidoses	GNPTA	E
Muscle phosphorylase	PYGM	E
N-acetylgalactosamine-6-sulfate sulfatase	GALNS	E
N-acetylglucosamine-6-sulfatase	GNS	E
N-acetylglucosaminidase, alpha	NAGLU	E
N-acetyltransferase 1	NAT1	E
N-acetyltransferase 2	NAT2	E
NADH dehydrogenase		E
NADH dehydrogenase (ubiquinone) Fe-S protein 1	NDUFS1	E
NADH dehydrogenase (ubiquinone) Fe-S protein 4	NDUFS4	E
NADH dehydrogenase (ubiquinone) flavoprotein 1	NDUFV1	E
NADH-cytochrome b5 reductase	DIA1	E
NADPH-dependent cytochrome P450 reductase	POR	E
Neuroendocrine convertase 1	NEC1, PCSK1	E
Neutral endopeptidase		E
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Nucleoside diphosphate kinase-A	NDPKA	E
Ornithine delta-aminotransferase	OAT	E
Ornithine transcarbamoylase	OTC, NME1	E
Pancreatic amylase		E
Pancreatic lipase	PNLIP	E
Pancreatic lipase related protein 1	PLRP1	E
Pancreatic lipase related protein 2	PLRP2	E

Paraoxonase PON1	PON1	E
Paraoxonase PON2	PON2	E
Paraoxonase PON3		E
PCNA (proliferating cell nuclear antigen)		E
Pepsinogen		E
Peroxidase, salivary	SAPX	E
Phenylalanine hydroxylase	PAH	E
Phenylalanine monooxygenase		E
Phenylethanolamine N-methyltransferase, PNMT	PNMT	E
Phosphoenolpyruvate carboxykinase	PCK1	E
Phosphofructokinase, liver	PFKL	E
Phosphofructokinase, muscle	PFKM	E
Phosphoglucomutase		E
Phosphoglucose isomerase	GPI	E
Phosphoglycerate kinase 1	PGK1	E
Phosphoglycerate mutase 2	PGAM2	E
Phosphoribosyl pyrophosphate synthetase	PRPS1	E
Phosphorylase kinase deficiency, liver	PHK	E
Phosphorylase kinase, alpha 1 (muscle)	PHKA1	E
Phosphorylase kinase, alpha 2	PHKA2	E
Phosphorylase kinase, beta	PHKB	E
Phosphorylase kinase, delta		E
Phosphorylase kinase, gamma 2	PHKG2	E
Pineolytic beta-receptors		E
Plasminogen	PLG	E
Plasminogen activator inhibitor 1	PAI1	E
Plasminogen activator inhibitor 2	PAI2	E
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E
Poly (ADP-ribose) synthetase	PARS	E
Porphobilinogen deaminase	HMBS	E
Procollagen N-protease		E
Procollagen peptidase		E
Proline dehydrogenase	PRODH	E
Prolyl-4-hydroxylase		E
Propionyl-CoA carboxylase, alpha	PCCA	E
Propionyl-CoA carboxylase, beta	PCCB	E
Prostasin, PRSS8	PRSS8	E
Protease nexin 2	PN2	E
Protective protein for beta-galactosidase	PPGB	E
Protein kinase A		E
Protein kinase B	PRKB	
Protein kinase C, alpha	PRKCA	E
Protein kinase C, gamma	PRKCG	E
Protein kinase DNA-activated	PRKDC	E
Protein kinase G		E
Protein phosphatase 1, regulatory (inhibitor) subunit 3	PPP1R3	E
Protein phosphatase 2, regulatory subunit A, beta	PPP2R1B	E

isoform		
Protoporphyrinogen oxidase	PPOX	E
Pterin-4-alpha-carbinolamine	PCBD	
Purine nucleoside phosphorylase	NP	E
Pyrroline-5-carboxylate synthetase	PYCS	E
Pyruvate carboxylase	PC	E
Pyruvate decarboxylase	PDHA	E
Pyruvate kinase	PKLR	E
Quinoid dihydropteridine reductase	QDPR	E
Renin	REN	E
Replication factor A		E
Replication factor C	RFC2	E
Rhodopsin kinase	RHOK	E
Ribonucleotide reductase, RRM		E
Ribosephosphate pyrophosphokinase		E
Ribosomal protein L13A	RPL13A	G
Ribosomal protein L17	RPL17	G
Ribosomal protein S19	RPS19	E
Ribosomal protein S4, X-linked	RPS4X	E
Ribosomal protein S6 kinase	RPS6KA3	E
Ribosomal protein S9	RPS9	G
S-adenosylmethionine decarboxylase, AMD		E
Serine hydroxymethyltransferase	SHMT	E
Serotonin N-acetyltransferase	SNAT	E
Sorbitol dehydrogenase	SORD	E
Sphingomyelinase	SMPD1	E
Steroid 5 alpha reductase 1	SRD5A1	E
Steroid 5 alpha reductase 2	SRD5A2	E
Steroid sulphatase	STS	E
Succinate dehydrogenase 1	SDH1	E
Succinate dehydrogenase 2	SDH2	E
Succinate thiokinase		E
Succinic semi-aldehyde dehydrogenase	ssadh	E
Succinyl CoA synthase		E
Sucrase		E
Sulfite oxidase	SUOX	E
Superoxide dismutase 1	SOD1	E
Superoxide dismutase 3	SOD3	E
TEK, tyrosine kinase, endothelial	TEK	E
Telomerase protein component		E
Terminal deoxynucleotidyltransferase, TDT		E
Thiolase, perioxisomal		E
Thiopurine S-methyltransferase	TPMT	E
Thymidylate synthase	TYMS	E
Tissue inhibitor of metalloproteinase 1, TIMP1	TIMP1	E
Tissue inhibitor of metalloproteinase 2, TIMP2	TIMP2	E
Tissue inhibitor of metalloproteinase 3, TIMP3	TIMP3	E
Tissue inhibitor of metalloproteinase 4, TIMP4	TIMP4	E
Tissue non-specific alkaline phosphatase TNSAP		E
Topoisomerase I		E

Fatty acid binding proteins FABP4		T
Fatty acid binding proteins FABP5		T
Fatty acid binding proteins FABP6		T
Ferritin, H subunit		T
Ferritin, L subunit	FTL	T
Fucosyltransferase 2	FUT2	T
Fucosyltransferase 3	FUT3	T
Fucosyltransferase 6	FUT6	T
Furin		T
Gamma-glutamyl carboxylase	GGCX	T
Gamma-glutamyltransferase 1	GGT1	T
Gamma-glutamyltransferase 2	GGT2	T
Gap junction protein alpha 1	GJA1	T
Gap junction protein alpha 3	GJA3	T
Gap junction protein alpha 8	GJA8	T
Gap junction protein beta 1	GJB1	T
Gap junction protein beta 2	GJB2	T
Gap junction protein beta 3	GJB3	T
Gastric inhibitory polypeptide GIP	GIP	T
Gastric inhibitory polypeptide receptor, GIPR	GIPR	T
Gastric lipase, LIPF		T
Gastrin releasing peptide	GRP	T
Gastrin releasing peptide receptor	GRPR	T
Glucagon synthase		T
Glutamine transporter		T
Glutathione	GSH	T
Guanylin	GUCA2	T
Haem oxygenase		T
Haemoglobin alpha 1	HBA1	T
Haemoglobin alpha 2	HBA2	T
Haemoglobin beta	HBB	T
Haemoglobin delta	HBD	T
Haemoglobin epsilon		T
Haemoglobin gamma A	HBG1	T
Haemoglobin gamma B	HBG2	T
Haemoglobin gamma G	HBGG	T
Hemochromatosis	HFE	T
Hermansky-pudlak syndrome gene	HPS	T
Histidine-rich glycoprotein	HRG	T
Huntingtin	HD	T
Hyaluronidase		T
Intestinal alkaline phosphatase IAP		T
Kell blood group precursor	XK, KEL	T
Lactotransferrin	LTF	T
Lipoprotein receptor, Low Density	LDLR	T
Lipoprotein, High Density	HDLDT1	T
Lipoprotein, Intermediate Density		T
Lipoprotein, Low Density 1		T
Lipoprotein, Low Density 2		T
Lipoprotein, Very Low Density	VLDLR	T

Long QT-type 2 potassium channels	LQT2, KCNH2	T
Low density lipoprotein receptor-related protein precursor	LRP	T
Mannosyl (alpha-1,6-)-glycoprotein beta-1, 2-N-acetylglucosaminyltransferase	MGAT2	T
Marenostrin	MEFV	T
Melanocortin 1 receptor	MC1R	T
Melanocortin 2 receptor	MC2R	T
Melanocortin 4 receptor	MC4R	T
Metallothionein		T
Microsomal triglyceride transfer protein	MTP	T
Mucin 18	MUC18	T
Mucin, MUC2		T
Mucin, MUC5AC		T
Mucin, MUC6		T
Mulibrey nanism	MUL	T
Myocilin	MYOC	T
Myoglobin		T
Myopia 1	MYP1	T
Myopia 2	MYP2	T
Na+/H+ exchanger 1	NHE1	T
Na+/H+ exchanger 2	NHE2	T
Na+/H+ exchanger 3	NHE3	T
Na+/H+ exchanger 4	NHE4	T
Na+/H+ exchanger 5	NHE5	T
Na+coupled glucose/galactose transporter		T
Nephrolithiasis 2	NPHL2	T
Nephronophthisis 1	NPHP1	T
Nephronophthisis 2	NPHP2	T
Nephrosis 1	NPHS1	T
Neuraminidase sialidase	NEU	T
Niemann-Pick disease protein	NPC1	T
Nucleophosmin	NPM1	T
Palmitoyl-protein thioesterase	PPT	T
Pancreatic colipase		T
Pendrin, PDS	PDS	T
Pepsin		T
Peptidases A		T
Peptidases B		T
Peptidases C		T
Peptidases D	PEPD	T
Peptidases E		T
Peptidases S		T
Peroxisomal membrane protein 3	PXMP3	T
Peroxisome biogenesis factor 1	PEX1	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome biogenesis factor 19	PEX19	T
Peroxisome proliferative activated receptor, alpha	PPARA	T
Peroxisome proliferative activated receptor, gamma	PPARG	T

Peroxisome receptor 1	PXR1	T
P-glycoprotein 1	PGY1	T
P-glycoprotein 3	PGY3	T
Phosphomannomutase-2	PMM2	T
Phosphomannose isomerase-1, PMI1	MPI	T
Plakophilin 1	PKP1	T
Platelet glutaminase	GLS	T
Platelet monamine oxidase		T
Plectin 1	PLEC1	T
Polycystic kidney and hepatic disease 1	PKHD1	T
Polycystin 1	PKD1	T
Polycystin 2	PKD2	T
Polymorphonuclear elastase		T
Preproglucagon		T
Preproinsulin		T
Presenilin 1	PSEN1	T
Presenilin 2	PSEN2	T
Prostaglandin I2 receptor		T
Protease inhibitor 1		T
Renal glutaminase		T
Retinaldehyde binding protein 1	RLBP1	T
Retinol binding protein 1		T
Retinol binding protein 2		T
Retinol binding protein 4	RBP4	T
Rhesus blood group, CcEe antigens	RHCE	T
Rhesus blood group, D antigen	RHD	T
Rhesus blood group-associated glycoprotein	RHAG	T
Salivary amylase, AMY1		T
Secretin	SCT	T
Secretin receptor, SCTR	SCTR	T
Serum amyloid A	SAA	T
Serum amyloid P	SAP	T
Sex hormone binding globulin, SHBG		T
Solute carrier family 1 (amino acid transporter), member 6	SLC1A6	T
Solute carrier family 1 (glial high affinity glutamate transporter), member 3	SLC1A3	T
Solute carrier family 1 (glutamate transporter), member 1	SLC1A1	T
Solute carrier family 1 (glutamate transporter), member 2	SLC1A2	T
Solute carrier family 1 (neutral amino acid transporter), member 4	SLC1A4	T
Solute carrier family 10 (sodium/bile acid cotransporter family),member 1	SLC10A1	T
Solute carrier family 10 (sodium/bile acid cotransporter family),member 2	SLC10A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T
Solute carrier family 12, member 3	SLC12A3	T

Solute carrier family 14, member 2	SLC14A2	T
Solute carrier family 15 (H+/peptide transporter, intestinal), member 1	SLC15A1	T
Solute carrier family 15 (H+/peptide transporter, kidney), member 2	SLC15A2	T
Solute carrier family 16 (monocarboxylate transporter), member 1	SLC16A1	T
Solute carrier family 16 (monocarboxylate transporter), member 7	SLC16A7	T
Solute carrier family 17, member 1	SLC17A1	T
Solute carrier family 17, member 2	SLC17A2	T
Solute carrier family 18, member 3	SLC18A3	T
Solute carrier family 19 (folate transporter), member 1	SLC19A1	T
Solute carrier family 2 (facilitated glucose transporter), member 1	SLC2A1	T
Solute carrier family 2 (facilitated glucose transporter), member 2	SLC2A2	T
Solute carrier family 2 (facilitated glucose transporter), member 3	SLC2A3	T
Solute carrier family 2 (facilitated glucose transporter), member 4	SLC2A4	T
Solute carrier family 2 (facilitated glucose transporter), member 5	SLC2A5	T
Solute carrier family 20, member 1	SLC20A1	T
Solute carrier family 20, member 2	SLC20A2	T
Solute carrier family 20, member 3	SLC20A3	T
Solute carrier family 21, member 2	SLC21A2	T
Solute carrier family 21, member 3	SLC21A3	T
Solute carrier family 22, member 1	SLC22A1	T
Solute carrier family 22, member 2	SLC22A2	T
Solute carrier family 22, member 5	SLC22A5	T
Solute carrier family 25, member 12	SLC25A12	T
Solute carrier family 3 (facilitated glucose transporter), member 1	SLC3A1	T
Solute carrier family 4 (anion exchanger), member 1	SLC4A1	T
Solute carrier family 4 (anion exchanger), member 2	SLC4A2	T
Solute carrier family 4 (anion exchanger), member 3	SLC4A3	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-AMINOBUTYRIC ACID transporter), member 1	SLC6A1	T

Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Solute carrier family 6, member 10	SLC6A10	T
Solute carrier family 6, member 6	SLC6A6	T
Solute carrier family 6, member 8	SLC6A8	T
Solute carrier family 7(amino acid transporter), member 1	SLC7A1	T
Solute carrier family 7(amino acid transporter), member 2	SLC7A2	T
Solute carrier family 7(amino acid transporter), member 7	SLC7A7	T
Solute carrier family 8 (sodium/calcium exchanger), member 1	SLC8A1	T
Sorcin	SRI	T
Steroidogenic acute regulatory protein	STAR	T
Sterol carrier protein 2	SCP2	T
Stratum corneum chymotryptic enzyme		T
Sucrase-isomaltase	SI	T
Surfactant pulmonary-associated protein A1	SFTPA1	T
Surfactant pulmonary-associated protein A2	SFTPA2	T
Surfactant pulmonary-associated protein B	SFTPB	T
Surfactant pulmonary-associated protein C	SFTPC	T
Surfactant pulmonary-associated protein D	SFTPD	T
Survival of motor neuron 1, telomeric	SMN1	T
Tetranectin	TNA	T
Thyroxin-binding globulin	TBG	T
Tocopherol (alpha) transfer protein	TTPA	T
Transcobalamin 1, TCN1		T
Transcobalamin 2, TCN2	TCN2	T
Transthyretin	TTR	T
Trehalase		T
Trypsinogen activation peptide		T
Uncoupling protein 1		T
Uncoupling protein 3	UCP3	T
Uteroglobin	UGB	T
Vitelliform macular dystrophy, atypical gene	VMD1	T
Vitronectin receptor, alpha	VNRA	T
Von Willebrand factor	VWF	T
Achromatopsia 2	ACHM2	S
Actin, alpha, skeletal	ACTA1	S
Actin, alpha, smooth, aortic	ACTA2	S
Actin, alpha, cardiac	ACTC	S
Actin, beta	ACTB	S
Actin, gamma 2	ACTG2	S
Adducin, alpha	ADD1	S
Adducin, beta	ADD2	S

Amelogenin	AMELX	S
Ankyrin 1	ANK1	S
Ankyrin 2	ANK2	S
Ankyrin 3	ANK3	S
Apaf-1		S
Arrestin	SAG	S
Blue cone pigment	BCP	S
Chloride channel 1, skeletal muscle	CLCN1	S
Chloride channel 5	CLCN5	S
Chloride channel KB	CLCNKB	S
Choroideremia gene	CHM	S
Cofilin		S
Collagen I alpha 1	COL1A1	S
Collagen I alpha 2	COL1A2	S
Collagen II alpha 1	COL2A1	S
Collagen III alpha 1	COL3A1	S
Collagen IV alpha 1	COL4A1	S
Collagen IV alpha 2	COL4A2	S
Collagen IV alpha 3	COL4A3	S
Collagen IV alpha 4	COL4A4	S
Collagen IV alpha 5	COL4A5	S
Collagen IV alpha 6	COL4A6	S
Collagen IX alpha 2	COL9A2, EDM2	S
Collagen IX alpha 3	COL9A3	S
Collagen receptor	COLR	S
Collagen V alpha 1	COL5A1	S
Collagen V alpha 2	COL5A2	S
Collagen VI alpha 1	COL6A1	S
Collagen VI alpha 2	COL6A2	S
Collagen VI alpha 3	COL6A3	S
Collagen VII alpha 1	COL7A1	S
Collagen X alpha 1	COL10A1	S
Collagen X alpha 1	COL11A1	S
Collagen XI alpha 2	COL11A2	S
Collagen XVII alpha 1	COL17A1	S
Cryptochrome 1	CRY1	S
Cryptochrome 2	CRY2	S
Crystallin, alpha A	CRYAA	S
Crystallin, alpha B	CRYAB	S
Crystallin, beta B2	CRYBB2	S
Crystallin, gamma A	CRYGA	S
Desmin	DES	S
DNA damage binding protein, DDB1	DDB1	S
DNA damage binding protein, DDB2	DDB2	S
DNA-damage-inducible transcript 3	DDIT3	S
Doublecortin, DCX	DCX	S
Dyskerin	DKC1	S
Dystonia 1	DYT1	S
Dystonia 3	DYT3	S
Dystonia 6	DYT6	S

Dystonia 7	DYT7	S
Dystonia 9	CSE	S
Dystrophin	DMD	S
Dystrophin-associated glycoprotein 35kD, SCGD	SGCD	S
Dystrophin-associated glycoprotein 35kD, SGSG	SGCG	S
Dystrophin-associated glycoprotein 43kD	SGCB	S
Dystrophin-associated glycoprotein 50kD	SGCA	S
Ectodermal Dysplasia 1 gene	ED1	S
Elastin	ELN	S
Endocardial fibroelastosis 2 gene	EFE2	S
Endoglin	ENG	S
Erythrocyte membrane protein band 4.1	EPB41	S
Erythrocyte membrane protein band 4.2	EPB42	S
Erythrocyte membrane protein band 7.2	EPB72	S
Exostosin 1	EXT1	S
Exostosin 2	EXT2	S
Exostosin 3	EXT3	S
Eye colour gene 3 (brown)	EYCL3	S
Fibrinogen alpha	FGA	S
Fibrinogen beta	FGB	S
Fibrinogen gamma	FGG	S
Glycophorin A	GYPA	S
Glycophorin B	GYPB	S
Glycophorin C	GYPC	S
Green cone pigment	GCP	S
Keratin 1	KRT1	S
Keratin 10	KRT10	S
Keratin 11	KRT11	S
Keratin 12	KRT12	S
Keratin 13	KRT13	S
Keratin 14	KRT14	S
Keratin 15	KRT15	S
Keratin 16	KRT16	S
Keratin 17	KRT17,PCHC1	S
Keratin 18	KRT18	S
Keratin 2	KRT2	S
Keratin 3	KRT3	S
Keratin 4	KRT4	S
Keratin 5	KRT5	S
Keratin 6	KRT6	S
Keratin 7	KRT7	S
Keratin 8	KRT8	S
Keratin 9	KRT9	S
Keratin, hair acidic 1	KRTHA1	S
Keratin, hair basic 2	KRTHB1	S
Keratin, hair basic 6	KRTHB6	S
Loricrin	LOR	S
Microtuble associated protein	MAP	S
Moesin, MSN		S
Myomesin 1	MYOM1	S

Myomesin 2	MYOM2	S
Myelin basic protein		S
Myelin protein peripheral 22	PMP22	S
Myelin protein zero	MPZ	S
Myosin 15	MYO15	S
Myosin 5A	MYO5A	S
Myosin 6	MYO6	S
Myosin 7A	MYO7A	S
Myosin, cardiac	MYH7	S
Myosin, light chain 2	MYL2	S
Myosin, light chain 3	MYL3	S
Myosin-binding protein C, cardiac	MYBPC3	S
Myotubularin	MTM1	S
Nebulin	NEB	S
Neurofilament protein, heavy	NFH	S
Neurofilament protein, NF125	NF150	S
Neurofilament protein, NF200	NF200	S
Neurofilament protein, NF68	NF68	S
Ocular albinism 1	OA1	S
Oculocutaneous albinism II	OCA2	S
Osteocalcin		S
Peripherin, PRPH		S
Peroxisomal membrane protein 1	PXMP1	S
Persyn		S
Proline-rich protein BstNI subfamily 1	PRB1	S
Proline-rich protein BstNI subfamily 3	PRB3	S
Proline-rich protein BstNI subfamily 4	PRB4	S
Radixin	RDX	S
Red cone pigment	RCP	S
Retinal pigment epithelium specific protein (65kD)	RPE65	S
Retinitis pigmentosa gene 1	RP1	S
Retinitis pigmentosa gene 2	RP2	S
Retinitis pigmentosa gene 3	RP3	S
Retinitis pigmentosa gene 6	RP6	S
Retinitis pigmentosa gene 7	RP7, RDS	S
Rhodopsin	RHO	S
Rod outer segment membrane protein 1	ROM1	S
Semaphorin A4	SEMA4	S
Semaphorin A5	SEMA5	S
Semaphorin D		S
Semaphorin E	SEMAE	S
Semaphorin F	SEMA3/F	S
Semaphorin W	SEMAW	S
Small nuclear ribonucleoprotein polypeptide N	SNRPN	S
Spectrin alpha	SPTA1	S
Spectrin beta	SPTB	S
Talin, TLN		S
Tau protein	MAPT	S
Tenascin (cytotactin)		S
Tenascin XA	TNXA	S

Titin	TTN	S
Tropomyosin 1 alpha	TPM1	S
Tropomyosin 3 (non-muscle)	TPM3	S
Troponin C		S
Troponin I	TNNI3	S
Troponin T2, cardiac	TNNT2	S
Tubulin		S
Undulin 1	COL14A1	S
Usher syndrome 2A	USH2A	S
Villin		S
Vinculin		S
Wolfram syndrome 1 gene	WFS1	S
Zinc finger protein 198	ZIC198	S
Zinc finger protein 2	ZIC2	S
Zinc finger protein 3	ZIC3	S
Zinc finger protein HRX	ALL1	I
Alpha 2 macroglobulin	A2M	I
Annexin 1	ANX 1	I
Apoptosis antigen 1	APT1	I
Apoptosis antigen ligand 1	APT1LG1	I
Apoptosis-inducing factor	AIF	I
ATP-binding cassette transporter 7	ABC7	I
Attractin		I
Autoimmune regulator, AIRE	AIRE	I
B-cell CLL/lymphoma 1	BCL1	I
B-cell CLL/lymphoma 10	BCL10	I
B-cell CLL/lymphoma 3	BCL3	I
B-cell CLL/lymphoma 4	BCL4	I
B-cell CLL/lymphoma 5	BCL5	I
B-cell CLL/lymphoma 6	BCL6	I
B-cell CLL/lymphoma 7	BCL7	I
B-cell CLL/lymphoma 8	BCL8	I
B-cell CLL/lymphoma 9	BCL9	I
beta 2 microglobulin	B2M	I
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Calcineurin A1	CALNA1	I
Calcineurin A2	CALNA2	I
Calcineurin A3	CALNA3	I
Calcineurin B		I
Catalase	CAT	I
CD1	CD1	I
CD10	CD10	I
CD100	CD100	I
CD101	CD101	I
CD103	CD103	I
CD106	CD106	I
CD107	CD107	I
CD108	CD108	I
CD109	CD109	I

CD110	I
CD111	I
CD112	I
CD113	I
CD114	I
CD115	I
CD116	I
CD117	I
CD118	I
CD119	I
CD12	I
CD120	I
CD121	I
CD122	I
CD123	I
CD124	I
CD125	I
CD126	I
CD127	I
CD128	I
CD129	I
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CD130	I
CD131	I
CD132	I
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CD140	I
CD141	I
CD142	I
CD143	I
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CD145	I
CD147	I
CD148	I
CD149	I
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CD152	I
CD153	I
CD154	I
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CD156	I

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CD158	I
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CD166	I
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CD2	I
CD20	I
CD22	I
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CD25	I
CD26	I
CD27	I
CD28	I
CD3	I
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CD31	I
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CD39	I
CD4	I
CD40	I
CD41	I
CD42	I
CD43	I
CD44	I
CD45	I
CD46	I
CD47	I
CD48	I
CD5	I
CD50	I
CD52	I
CD53	I
CD55	I
CD57	I
CD58	I
CD59	I
CD6	I
CD60	I

CD63	CD63	I
CD65	CD65	I
CD66	CD66	I
CD67	CD67	I
CD68	CD68	I
CD69	CD69	I
CD7	CD7	I
CD70	CD70	I
CD71	CD71	I
CD72	CD72	I
CD73	CD73	I
CD74	CD74	I
CD75	CD75	I
CD76	CD76	I
CD77	CD77	I
CD78	CD78	I
CD79	CD79	I
CD8	CD8	I
CD80	CD80	I
CD81	CD81	I
CD83	CD83	I
CD84	CD84	I
CD85	CD85	I
CD86	CD86	I
CD88	CD88	I
CD89	CD89	I
CD9	CD9	I
CD90	CD90	I
CD91	CD91	I
CD92	CD92	I
CD93	CD93	I
CD94	CD94	I
CD96	CD96	I
CD97	CD97	I
CD98	CD98	I
CD99	CD99	I
Chemokine MCAF	MCAF	I
Chemokine receptor CCR2	CCR2	I
Chemokine receptor CCR3	CCR3	I
Chemokine receptor CCR5	CCR5	I
Chemokine receptor CXCR1	CXCR1	I
Chemokine receptor CXCR2	CXCR2	I
Chemokine receptor CXCR4	CXCR4	I
Cholesterylester hydrolase		I
Chondritin Sulphate A - placental receptor		I
Cochlin	COCH	I
Complement component C1 inhibitor	C1NH	I
Complement component C1qa	C1QA	I
Complement component C1qb	C1QB	I
Complement component C1qg	C1QG	I

Complement component C1r	C1R	I
Complement component C1s	C1S	I
Complement component C2	C2	I
Complement component C3	C3	I
Complement component C4A	C4A	I
Complement component C4B	C4B	I
Complement component C5	C5	I
Complement component C6	C6	I
Complement component C7	C7	I
Complement component C8	C8B	I
Complement component C9	C9	I
Complement component receptor 1	CR1	I
Complement component receptor 2	CR2	I
Complement component receptor 3	CR3	I
Corticosteroid nuclear receptor		I
Cortisol receptor		I
C-reactive protein CRP		I
Cyclophilin		I
Cytokine-suppressive antiinflammatory drug-binding protein 1	CSBP1	I
Cytokine-suppressive antiinflammatory drug-binding protein 2	CSBP2	I
DAX1 nuclear receptor	DAX1	I
Endo-P-D-glucuronidase		I
Erythropoietin	EPO	I
Erythropoietin receptor	EPOR	I
Factor 1 (No. one)	F1	I
Factor B, properdin		I
Factor D		I
Factor H	HF1	I
Factor I (letter I)	IF	I
Factor III	F3	I
Factor IX	F9	I
Factor V	F5	I
Factor VII	F7	I
Factor VIII	F8	I
Factor X	F10	I
Factor XI	F11	I
Factor XII	F12	I
Factor XIII A & B	F13A & F13B	I
Fc receptor		I
Follicular lymphoma variant translocation 1	FVT1	I
Gastrointestinal tumor-associated antigen 1	GA733	I
Growth-regulated protein precursor, GRO	GRO	I
Haptoglobin, alpha 1	HPA1	I
Haptoglobin, alpha 2	HPA2	I
Haptoglobin, beta	HPB	I
Heat shock protein, HSP60		I
Heat shock protein, HSP70		I
Heat shock protein, HSP90		I

Heat shock protein, HSPA1		I
Heat shock protein, HSPA2		I
Hemopexin	HPX	I
Heparin Cofactor II	HCF2	I
Hepatitis B virus integration site 1	HVBS1	I
Hepatitis B virus integration site 2	HVBS6	I
Histatin 1		I
Histatin 2		I
Histatin 3	HTN3	I
HLA-B associated transcript 1	BAT1	I
IC7 A and B		I
Immunoglobulin alpha (IgA)	IGHA	I
Immunoglobulin gamma (IgG) 2	IGHG2	I
Immunoglobulin delta (IgD)	IGHD	I
Immunoglobulin epsilon (IgE)	IGHE	I
Immunoglobulin E (IgE) reponsiveness gene	IGER	I
Immunoglobulin E (IgE) serum concentration regulator gene	IGES	I
Immunoglobulin heavy mu chain	IGHM	I
Immunoglobulin J polypeptide	IGJ	I
Immunoglobulin kappa constant region	IGKC	I
Immunoglobulin kappa variable region	IGKV	I
Intercellular adhesion molecule 1	ICAM1	I
Intercellular adhesion molecule 2	ICAM2	I
Intercellular adhesion molecule 3	ICAM3	I
Interferon alpha	IFNA1	I
Interferon beta	IFNB	I
Interferon gamma	IFNG	I
Interferon gamma receptor 1	IFNGR1	I
Interferon gamma receptor 2	IFNGR2	I
Interferon regulatory factor 1	IRF1	I
Interferon regulatory factor 4	IRF4	I
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I
Interleukin(IL) 4 receptor	IL4R	I

Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
Kallikrein 3	KAK3	I
Kininogen, High molecular weight	KNG	I
Lectin, mannose-binding 1	LMAN1	I
Lectin, mannose-binding 2	MBL2	I
Leukin		I
Leukocyte-specific transcript 1	LST-1	I
Leukotriene A4 hydrolase		I
Leukotriene B4 receptor		I
Leukotriene C4 receptor		I
Leukotriene D4/E4 receptor		I
LIM-Kinase I (LINK-I)		I
Lipocortin 1	ANX4	I
Lipoprotein lipase	LPL	I
Lipoprotein-associated coagulation factor	LACI	I
Lipoxygenase 12 (platelets)	LOG12	I
Lipoxygenase 5 (leukocytes)		I
Lymphoblastic leukemia derived sequence 1	LYL1	I
Lymphocyte-specific protein tyrosine kinase	LCK	I
lymphotoxin		I
Lysozyme	LYZ	I
Macrophage activating factor	MAF	I
Macrophage inflammatory protein-1	MIP1	I
Macrophage inflammatory protein-1 receptor		I
Macrophage inflammatory protein-2	MIP2	I
Macrophage inflammatory protein-2 receptor		I
Malignant proliferation, eosinophil gene	MPE	I
Mannose binding protein	MBP	I
MHC Class I: A		I
MHC Class I: B		I
MHC Class I: C		I
MHC Class I: LMP-2, LMP-7		I
MHC Class I: Tap1	ABCR, TAP1	I
MHC Class II: DP	HLA-DPB1	I
MHC Class II: DQ		I
MHC Class II: DR		I
MHC Class II: Tap2	TAP2, PSF2	I
MHC Class II:Complementation group A	MHC2TA	I
MHC Class II:Complementation group B	rfxank	I
MHC Class II:Complementation group C	RFX5	I

MHC Class II:Complementation group D	RFXAP	I
Monocyte chemoattractant protein 1	MCP1	I
Myeloid leukemia factor-1	MLF1	I
Myeloperoxidase	MPO	I
N-acyl hydrolase		I
NADPH oxidase		I
Natural resistance-associated macrophage protein 1	NRAMP1	I
NB6		I
Neuronal apoptosis inhibitory protein	NAIP	I
Neuronal molecule-1		I
Neuronal molecule-1 receptor		I
Neutrophil cystolic factor 1	NCF1	I
Neutrophil cystolic factor 2	NCF2	I
Nuclear factor I-kappa-B-like gene	IKBL	I
Nuclear factor kappa beta	NFKB	I
Peanut-like 1	PNUTL1	I
Phagocytin		I
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Platelet glycoprotein 1b, alpha	GP1BA	I
Platelet glycoprotein 1b, beta	GP1BB	I
Platelet glycoprotein 1b, gamma	GP1BG	I
Platelet glycoprotein IX	GP9	I
Platelet glycoprotein V	GP5	I
Platelet-activating factor acetylhydrolase 1B	PAFAH1B1 or LIS1	I
Platelet-activating factor acetylhydrolase 2	PAFAH2	I
Platelet-activating factor receptor	PAFR	I
Poliovirus receptor	PVR, PVS	I
Prekallikrein		I
Properdin P factor, complement	PFC, PFD	I
Prostacyclin synthase		I
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin F2 alpha receptor		I
Prostaglandin IP receptor		I

Protein C	PROC	I
Protein C inhibitor	PCI	I
Protein S	PROS1	I
Proteinase 3		I
Prothrombin precursor	F2	I
SAP (SLAM-associated protein)	SH2D1A	I
Severe combined immunodeficiency, type A (Athabascan)	SCIDA	I
Signaling lymphocyte activation molecule	SLAM	I
Sjoegren (Sjogren) syndrome antigen A1	SSA1	I
SYK-related tyrosine kinase	SRK	I
T-cell acute lymphocytic leukemia 1	TAL1	I
T-cell acute lymphocytic leukemia 2	TAL2	I
T-cell receptor, alpha	TCRA	I
T-cell receptor, delta	TCRD	I
Terminal deoxynucleotidyltransferase	TDT	I
Thrombin receptor	F2R	I
Thrombomodulin	THBD	I
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thy-1 T-cell antigen	THY1	I
Thymic humoral factor		I
Thymosin		I
Tip-associated protein	TAP	I
Toll-like receptor 4	TLR4	I
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I
Tumour suppresssor gene DRA	DRA	I
Uridine monophosphate kinase	UMPK	I
Uridine monophosphate synthetase	UMPS	I
Vimentin	VIM	I
Wiskott-Aldrich syndrome protein	WASP, THC	I
17-ketosteroid reductase		N
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N
Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N

Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N
Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N
Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNB1	N
Acetylcholine receptor, nicotinic, beta 2	CHRNB2	N
Acetylcholine receptor, nicotinic, beta 3	CHRNB3	N
Acetylcholine receptor, nicotinic, beta 4	CHRNB4	N
Acetylcholine receptor, nicotinic, epsilon	CHRNE	N
Acetylcholine receptor, nicotinic, gamma	CHRNG	N
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenyl cyclase		N
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
alpha thalassemia gene	ATRX	N
alpha-synuclein	SNCA	N
Amyloid beta (A4) precursor protein-binding, APBB1	APBB1	N
Amyloid beta A4 precursor protein	APP	N
Amyloid beta A4 precursor-like protein	APLP	N
Arginine vasopressin	AVP	N
Arginine vasopressin receptor 1A	AVPR1A	N
Arginine vasopressin receptor 1B	AVPR1B	N
Arginine vasopressin receptor 2	AVPR2	N
Aspartate receptor		N
Benzodiazepine receptor		N
beta-endorphin receptor		N
beta-synuclein	SNCB	N
Calcitonin receptor /Calcitonin gene-related peptide receptor	CALCR	N
Calcitonin/Calcitonin gene-related peptide alpha	CALCA	N
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N
Calcium channel, voltage-dependent, L type, alpha	CACNA1S	N

1S subunit		
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calcium channel, voltage-dependent, T-type		N
Calretinin	CALB2	N
Cannabinoid receptor	CNR1	N
Carnosinase		N
Cartilage oligomeric matrix protein	COMP, EDM1, PSACH	N
Cartilage-hair hypoplasia gene	CHH	N
Cellubrevin	CEB	N
Ceroid lipofuscinosis neuronal 2	CLN2	N
Ceroid lipofuscinosis neuronal 3	CLN3	N
Ceroid lipofuscinosis neuronal 4	CLN4	N
Ceroid lipofuscinosis neuronal 5	CLN5	N
Ceroid lipofuscinosis neuronal 6	CLN6	N
Cholecystokinin	CCK	N
Cholecystokinin B receptor	CCKBR	N
Corticosteroid binding globulin	CBG	N
Cyclic nucleotide gated channel alpha 1, CNGA1	CNGA1	N
Cyclic nucleotide gated channel alpha 3, CNGA3	CNGA3	N
Cystic fibrosis transmembrane conductance regulator, CFTR	CFTR	N
Deafness autosomal dominant 5	DFNA5	N
Deafness dystonia peptide	DDP	N
Diaphanous 1	DIAPH1	N
Diaphanous 2	DIAPH2	N
Dihydrolipoamide branched chain transacylase	DBT	N
Dihydrolipoamide dehydrogenase	DLD	N
Dihydrolipoamide succinyltransferase		N
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Dynorphin receptor		N
Endobrevin	VAMP8	N
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Fragile site, folic acid type, rare, fra(X) A	FRAXA	N
Fragile site, folic acid type, rare, fra(X) E	FRAXE	N
Fragile site, folic acid type, rare, fra(X) F	FRAXF	N
GABA receptor, alpha 1	GABRA1	N
GABA receptor, alpha 2	GABRA2	N

GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N
GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
Galanin	GAL	N
Galanin receptor	GALNR1	N
Gephyrin		N
Glial-cell derived neurotrophic factor (GDNF) receptor		N
Glial-cell derived neurotrophic factor, GDNF	GDNF	N
Glutamate receptor 1	GLUR1	N
Glutamate receptor 2	GLUR2	N
Glutamate receptor 3	GLUR3	N
Glutamate receptor 4	GLUR4	N
Glutamate receptor 5	GLUR5	N
Glutamate receptor 6	GLUR6	N
Glutamate receptor 7	GLUR7	N
Glutamate receptor, ionotropic, NMDA 1	NMDAR1	N
Glutamate receptor, ionotropic, NMDA 2A	NMDAR2A	N
Glutamate receptor, ionotropic, NMDA 2B	NMDAR2B	N
Glutamate receptor, ionotropic, NMDA 2C	NMDAR2C	N
Glutamate receptor, ionotropic, NMDA 2D	NMDAR2D	N
Glycine receptor, alpha	GLRA2	N
Glycine receptor, beta		N
Glycine transporter	GLYT	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 1, GNAI1	GNAI1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 2, GNAI2	GNAI2	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 3, GNAI3	GNAI3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS1	GNAS1	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS2	GNAS2	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS3	GNAS3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS4	GNAS4	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT1	GNAT1	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT2	GNAT2	N
Guanine nucleotide-binding protein, alpha	GNAO1	N

activating activity polypeptide, GNAO		
Guanine nucleotide-binding protein, beta polypeptide 3	GNB3	N
Guanine nucleotide-binding protein, gamma polypeptide 5	GNG5	N
Guanine nucleotide-binding protein, q polypeptide	GNAQ	N
Gustducin, alpha (taste-specific G protein)	GDCA	N
H(+), K(+) - ATPase	ATP4B	N
Hippocampal cholinergic neurostimulating peptide,	HCNP	N
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N
Inositol monophosphatase	IMPA1	N
Inositol polyphosphate 1-phosphatase	INPP1	N
Islet amyloid polypeptide	IAPP	N
L1 cell adhesion molecule	L1CAM	N
Luteinizing hormone-releasing hormone		N
Luteinizing hormone-releasing hormone receptor		N
Melatonin receptor 1A	MTNR1A	N
Melatonin receptor 1B	MTNR1B	N
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Neurexin		N
Neurite growth-promoting factor 2	MDK	N
Neurite inhibitory protein		N
Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Neurotensin	NTS	N
Neurotensin receptor	NTSR1	N
Opioid receptor, delta	OPRD1	N
Opioid receptor, kappa	OPRK1	N
Opioid receptor, mu	OPRM1	N
Otoferlin	OTOF	N
Oxytocin	OXT	N
Oxytocin receptor	OXTR	N
Parkin	PARK2	N
Pituitary adenylate cyclase activating peptide	PACAP	N
Pituitary adenylate cyclase activating peptide receptor	PACAP1R	N
Postsynaptic density-95 protein	PSD95	N
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium inwardly-rectifying channel J11	KCNJ11	N
Potassium voltage-gated channel A1	KCNA1	N
Potassium voltage-gated channel E1	KCNE1	N

Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
Potassium voltage-gated channel Q4	KCNQ4	N
Potassium channel, subfamily K, member 1	KCNK1	N
Potassium channel, subfamily K, member 2	KCNK2	N
Potassium channel, subfamily K, member 3	KCNK3	N
Potassium channel, calcium-activated,	KCNN4	N
Preproenkephalin	PENK	N
Prion protein	PRNP	N
Prodynorphin		N
Proopiomelanocortin	POMC	N
Prosaposin	PSAP	N
Proteolipid protein	PLP	N
Purinergic receptor P1A1		N
Purinergic receptor P1A2		N
Purinergic receptor P1A3		N
Purinergic receptor P2X, 1	P2RX1	N
Purinergic receptor P2X, 2	P2RX2	N
Purinergic receptor P2X, 3	P2RX3	N
Purinergic receptor P2X, 4	P2RX4	N
Purinergic receptor P2X, 5	P2RX5	N
Purinergic receptor P2X, 6	P2RX6	N
Purinergic receptor P2X, 7	P2RX7	N
Purinergic receptor P2Y, 1	P2RY1	N
Purinergic receptor P2Y, 2	P2RY2	N
Purinergic receptor P2Y, 11	P2RY11	N
Rabphilin		N
RAS-associated protein, RAB3A	RAB3A	N
Rim		N
S100 calcium-binding protein A1	S100A1	N
S100 calcium-binding protein A2	S100A2	N
S100 calcium-binding protein A3	S100A3	N
S100 calcium-binding protein A4	S100A4	N
S100 calcium-binding protein A5	S100A5	N
S100 calcium-binding protein A6	S100A6	N
S100 calcium-binding protein A7	S100A7	N
S100 calcium-binding protein A8	S100A8	N
S100 calcium-binding protein A9	S100A9	N
S100 calcium-binding protein B	S100B	N
S100 calcium-binding protein P	S100P	N
Secretase, alpha		N
Secretase, beta		N
Secretase, gamma		N
Selectin E	SELE	N
Selectin L	SELL	N
Selectin P	SELP	N
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N
Serotonin receptor, 5HT1C	HTR1C	N

Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage gated, type IV, alpha polypeptide	SCN4A	N
Sodium channel, voltage gated, type V, alpha polypeptide	SCN5A	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Somatostatin	SST	N
Somatostatin receptor, SSTR1	SSTR1	N
Somatostatin receptor, SSTR2	SSTR2	G
Somatostatin receptor, SSTR3	SSTR3	N
Somatostatin receptor, SSTR4	SSTR4	N
Somatostatin receptor, SSTR5	SSTR5	N
Spinocerebellar ataxia 8 gene	SCA8	N
Substance P		N
Synapsin 1a & 1b	SYN1	N
Synapsin 2a & 2b	SYN2	N
Synaptic vesicle amine transporter	SVAT	N
Synaptic vesicle protein 2	SV2	N
Synaptobrevin 1	SYB1	N
Synaptobrevin 2	SYB2	N
Synaptogyrin		N
Synaptophysin	SYP	N
Synaptosomal-associated protein, 25KD	SNAP25	N
Synaptotagmin 1	SYT1	N
Synaptotagmin 2	SYT2	N
Syntaxin 1	STX1	N
Tachykinin receptor, NK1R	TACR1	N
Tachykinin receptor, NK2R	TACR2	N
Tachykinin receptor, NK3R	TACR3	N
Thyrotropin releasing hormone	TRH	N
Thyrotropin releasing hormone receptor	TRHR	N
Transcription factor, TUPLE1	TUPLE1	N
Tremor, essential 1	ETM1	N
Tremor, essential 2	ETM2	N
Tryptophan 2,3-dioxygenase	TDO2	N
Vacuolar proton pump, subunit 1	VPP1	N

Vacuolar proton pump, subunit 3	VPP3	N
Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N
Vesicular monoamine transporter 1	VMAT1	N
Vesicular monoamine transporter 2	VMAT2	N
Absent in melanoma 1 gene	AIM1	G
Acrosin	ACR	G
Activin		G
Activin A receptor, type 2-like kinase 1	ACVRL1	G
Activin A receptor, type 2B	ACVR2B	G
Adenomatous polyposis coli tumour supressor gene	APC	G
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Aldosterone receptor	MLR	G
Alkaptonuria gene	AKU	G
alpha tectorin	TECTA	G
alpha-actinin 2	ACTN2	G
alpha-actinin 3	ACTN3	G
Alpha-fetoprotein	AFP	G
Amphiregulin	AREG	G
Androgen receptor	AR	G
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Anti-Mullerian hormone	AMH	G
Anti-Mullerian hormone type 2 receptor	AMHR2	G
AP-2, alpha	TFAP2A	G
AP-2, beta	TFAP2B	G
AP-2, gamma	TFAP2C	G
Apical protein, xenopus laevis-like	APXL	G
Apopain	CPP32	G
Archaete-scute homolog 1	ASH1	G
Archaete-scute homolog 2	ASH2	G
Astrotactin	ASTN	G
Ataxia telangiectasia complementation group D	ATD, ATDC	G
Ataxia telangiectasia gene, AT	ATM	G
Ataxin 1	SCA1	G
Ataxin 2	SCA2	G
Ataxin 3	MJD	G
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Atrophin 1	DRPLA	G
Azoospermia factor 1	AZF1	G
Bagpipe homeobox, drosophila homolog of, 1	BAPX1	G
BCL2-associated X protein	BAX	G
BCL2-related protein A1	BCL2A1	G
Beckwith-Wiedemann region 1A	BWR1A	G
Bloom syndrome protein	BLM	G
Bone morphogenetic protein, BMP1	BMP1	G
Bone morphogenetic protein, BMP2	BMP2	G

Cyclin-dependent kinase 9	CDK9	G
Cyclin-dependent kinase inhibitor 1A (P21, CIP1)	CDKN1A	G
Cyclin-dependent kinase inhibitor 1B (P27, KIP1)	CDKN1B	G
Cyclin-dependent kinase inhibitor 1C (P57, KIP2)	CDKN1C	G
Cyclin-dependent kinase inhibitor 2A (p16)	CDKN2A	G
Cyclin-dependent kinase inhibitor 3	CDKN3	G
Defender against cell death 1	DAD1	G
Deleted in azoospermia	DAZ	G
Deleted in colorectal carcinoma	DCC	G
Deleted in malignant brain tumours 1	DMBT1	G
Dentin sialophosphoprotein	DSPP	G
Desert hedgehog, dhh		G
Disrupted meiotic cDNA 1, homolog	DMC1	G
Distal-less homeobox 1	DLX1	G
Distal-less homeobox 2	DLX2	G
Distal-less homeobox 3	DLX3	G
Distal-less homeobox 4	DLX4	G
Distal-less homeobox 5	DLX5	G
Distal-less homeobox 6	DLX6	G
Dynamin	DNM1	G
Dynein		G
E74-like factor 1, ELF1	ELF1	G
EB1		G
Empty spiracles (drosophila) homologue 1	EMX1	G
Empty spiracles (drosophila) homologue 2	EMX2	G
Endometrial bleeding-associated factor	EBAF	G
Engrailed-1	EN1	G
Engrailed-2	EN2	G
Ephrin receptor tyrosine kinase A	EPHA	G
Ephrin receptor tyrosine kinase B	EPHB	G
Ephrin-A	EFNA	G
Ephrin-B	EFNB	G
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Erythroid kruppel-like factor	EKLF	G
Estrogen receptor	ESR	G
Eukaryotic initiation translation factor	EIF4E	G
EWS RNA-binding protein	EWSR1	G
Eyes absent 1	EYA1	G
Eyes absent 2	EYA2	G
Eyes absent 3	EYA3	G
Fc fragment of IgG, high affinity IA, receptor for	FCGR1A	G
Fc fragment of IgG, low affinity IIa, receptor for (CD32)	FCGR2A	G
Fc fragment of IgG, low affinity IIIa, receptor for (CD16)	FCGR3A	G
Fertilin protein	FTNB	G
Fibrillin 1	FBN1	G
Fibrillin 2	FBN2	G
Fibroblast growth factor	FGF1	G

Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G
Fibroblast growth factor receptor 3	FGFR3	G
Fibronectin precursor	FN1	G
Flightless-II, Drosophila homolog of	FLII	G
Folic acid receptor	FOLR	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Follistatin		G
Forkhead rhabdomyosarcoma gene	FKHR	G
Forkhead transcription factor 10	FKHL10	G
Forkhead transcription factor 14	FKHL14	G
Forkhead transcription factor 7	FKHL7	G
Frataxin	FRDA	G
Fringe secreted protein, lunatic	LFNG	G
Fringe secreted protein, manic	MFNG	G
Fringe secreted protein, radical	RFNG	G
Fukuyama type congenital muscular dystrophy	FCMD	G
G/T mismatch binding protein	GTBP, MSH6	G
Galactosyltransferase 1	GT1	G
Galactosyltransferase, alpha 1,3	GGTA1	G
Galactosyltransferase, beta 3	B3GALT	G
Gastrin	GAS	G
Gastrulation brain homeobox 2	GBX2	G
GDP dissociation inhibitor 1	GDI1	G
Gelsolin	GSN	G
Geniospasm 1	GSM1	G
Glioma chloride ion channel, GCC		G
Glucagon receptor	GCGR	G
Glucagon-like peptide receptor 1	GLP1R	G
Glucocorticoid receptor	GRL	G
Glypican 3	GPC3, SDYS	G
Gonadotropin releasing hormone	GNRH	G
Gonadotropin releasing hormone receptor	GNRHR	G
Goosecoid GSC		G
Growth arrest-specific homeobox	GAX	G
Growth factor receptor-bound protein 2	GRB2	G
Growth hormone 1	GH1	G
Growth hormone 2 (placental)	GH2	G
Growth hormone receptor	GHR	G
Growth hormone releasing hormone (GHRH)	GHRH	G
Growth hormone releasing hormone receptor	GHRHR	G
Growth/differentiation factor 5	GDF5	G
GTP cylcohydrolase 1	GCH1	G
GTPase-activating protein, GAP	RASA1	G
Hairless	HR	G
Hela tumor suppression gene	HTS1	G
Heparin binding epidermal growth factor	HBEGF	G
Hepatocyte growth factor	HGF	G
High mobility group protein 1	HMG1	G

High mobility group protein 2	HMG2	G
High mobility group protein C	HMGIC	G
High mobility group protein Y	HMGIY	G
Histone family H1	H1	G
Histone family H2	H2	G
Histone family H3	H3	G
Histone family H4	H4	G
HLH transcription factor HAND1	HAND1	G
HLH transcription factor HAND2	HAND2	G
Holoprosencephaly 1	HPE1	G
Holoprosencephaly 2	HPE2	G
Holoprosencephaly 3	HPE3	G
Holoprosencephaly 4	HPE4	G
Homeobox (HOX) gene A1	HOXA1	G
Homeobox (HOX) gene A2	HOXA2	G
Homeobox (HOX) gene A3	HOXA3	G
Homeobox (HOX) gene A4	HOXA4	G
Homeobox (HOX) gene A5	HOXA5	G
Homeobox (HOX) gene A6	HOXA6	G
Homeobox (HOX) gene A7	HOXA7	G
Homeobox (HOX) gene A8	HOXA8	G
Homeobox (HOX) gene A9	HOXA9	G
Homeobox (HOX) gene A10	HOXA10	G
Homeobox (HOX) gene A11	HOXA11	G
Homeobox (HOX) gene A12	HOXA12	G
Homeobox (HOX) gene A13	HOXA13	G
Homeobox (HOX) gene B1	HOXB1	G
Homeobox (HOX) gene B2	HOXB2	G
Homeobox (HOX) gene B3	HOXB3	G
Homeobox (HOX) gene B4	HOXB4	G
Homeobox (HOX) gene B5	HOXB5	G
Homeobox (HOX) gene B6	HOXB6	G
Homeobox (HOX) gene B7	HOXB7	G
Homeobox (HOX) gene B8	HOXB8	G
Homeobox (HOX) gene B9	HOXB9	G
Homeobox (HOX) gene C4	HOXC4	G
Homeobox (HOX) gene C8	HOXC8	G
Homeobox (HOX) gene C9	HOXC9	G
Homeobox (HOX) gene C13	HOXC13	G
Homeobox (HOX) gene D1	HOXD1	G
Homeobox (HOX) gene D3	HOXD3	G
Homeobox (HOX) gene D4	HOXD4	G
Homeobox (HOX) gene D8	HOXD8	G
Homeobox (HOX) gene D9	HOXD9	G
Homeobox (HOX) gene D10	HOXD10	G
Homeobox (HOX) gene D12	HOXD12	G
Homeobox (HOX) gene D13	HOXD13	G
Homeobox 11	HOX11	G
Homeobox HB24	HLX1	G
Homeobox HB9	HLXB9	G

Homeobox, PROX1	PROX1	G
Human atonal gene	ATOH1	G
Human chorionic gonadtrophin, hCG	CG	G
Human placental lactogen	CSH1	G
Ikaros gene	IKAROS	G
Indian hedgehog, ihh	IHH	G
Inhibin, alpha	INHA	G
Inhibin, beta A	INHBA	G
Inhibin, beta B	INHBB	G
Inhibin, beta C	INHBC	G
Inositol 1,4,5-triphosphate receptor 1	ITPR1	G
Inositol 1,4,5-triphosphate receptor 3	ITPR3	G
Insulin	INS	G
Insulin promotor factor 1	IPF1	G
Insulin receptor	INSR	G
Insulin receptor substrate-1	IRS1	G
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Integrin beta 3	ITGB3	G
Integrin beta 4	ITGB4	G
Integrin beta 5	ITGB5	G
Integrin beta 6	ITGB6	G
Integrin beta 7	ITGB7	G
Integrin, alpha 1	ITGA1	G
Integrin, alpha 2	ITGA2	G
Integrin, alpha 3	ITGA3	G
Integrin, alpha 4	ITGA4	G
Integrin, alpha 5	ITGA5	G
Integrin, alpha 6	ITGA6	G
Integrin, alpha 7	ITGA7	G
Integrin, alpha 8	ITGA8	G
Integrin, alpha 9	ITGA9	G
Integrin, alpha M	ITGAM	G
Integrin, alpha X	ITGAX	G
Janus kinase 1	JAK1	G
Janus kinase 2	JAK2	G
Janus kinase 3	JAK3	G
Kallman syndrome gene 1	KAL1	G
Kinectin	KTN1	G
Kinesin, heavy chain	KNSL1	G
Kinesin, light chain	KNS2	G
Lamin A/C	LMNA	G
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G
Laminin M	LAMM	G

Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukaemia inhibitory factor	LIF	G
Leukaemia inhibitory factor receptor	LIFR	G
LH/choriogonadotropin (CG) receptor	LHCGR	G
LIM homeobox protein 1	LHX1	G
LIM homeobox protein 2	LHX2	G
LIM homeobox protein 3	LHX3	G
LIM homeobox protein 4	LHX4	G
LIM homeobox transcription factor 1, beta	LMX1B	G
Limb girdle muscular dystrophy 1A	LGMD1A	G
Limb girdle muscular dystrophy 1B	LGMD1B	G
Limb girdle muscular dystrophy 2G	LGMD2G	G
Limb girdle muscular dystrophy 2H	LGMD2H	G
Limbic associated membrane protein	LAMP	G
LIM-domain only protein 1	LMO1	G
LIM-domain only protein 2	LMO2	G
LIM-domain only protein 3	LMO3	G
LIM-domain only protein 4	LMO4	G
Lipoma-preferred partner gene	LPP	G
Luteinizing hormone, beta chain	LHB	G
Lymphoid enhancer-binding factor	LEF-1	G
Lysosome-associated membrane protein 1	LAMP1	G
Lysosome-associated membrane protein 2	LAMP2	G
MAD (mothers against decapentaplegic, Drosophila) homologue 2	MADH2	G
MAD (mothers against decapentaplegic, Drosophila) homologue 3	MADH3	G
MAD (mothers against decapentaplegic, Drosophila) homologue 4	MADH4	G
MADS box transcription-enhancer factor 2A	MEF2A	G
MADS box transcription-enhancer factor 2B	MEF2B	G
MADS box transcription-enhancer factor 2C	MEF2C	G
MADS box transcription-enhancer factor 2D	MEF2D	G
MAPK kinase 1	MAPKK1; MEK1	G
MAPK kinase 4	MAPKK4; MEK4; SERK1	G
MAPK kinase 6	MAPKK6; MEK6	G
MAPKK kinase	MAPKKK	G
Matrix Gla protein	MGP	G
MAX-interacting protein 1	MXI1	G
Menin	MEN1	G
Mesoderm-specific transcript	MEST	G
Microphthalmia-associated transcription factor	MITF	G
Midline 1	MID1	G
Mismatch repair gene, PMSL1	PMS1	G
Mismatch repair gene, PMSL2	PMS2	G

Mitogen-activated protein (MAP) kinase	MAPK	G
Motilin	MLN	G
Msh homeobox homolog 1	MSX1	G
Msh homeobox homolog 2	MSX2	G
Multidrug resistance associated protein	MRP	G
Mutated in colorectal cancers, MCC	MCC	G
MutL homolog 1	MLH1	G
MutS homolog 2	MSH2	G
MutS homolog 3	MSH3	G
Myelodysplasia syndrome 1 gene	MDS1	G
Myogenic factor 3	MYF3	G
Myogenic factor 4	MYF4	G
Myogenic factor 5	MYF5	G
Na+, K+ ATPase, alpha	ATP1A1	G
Na+, K+ ATPase, beta 1	ATP1B1	G
Na+, K+ ATPase, beta 2	ATP1B2	G
Na+, K+ ATPase, beta 3	ATP1B3	G
Necdin	NDN	G
Nerve growth factor	NGF	G
Nerve growth factor receptor	NGFR	G
Neural retina-specific gene	NRL	G
Neuregulin	HGL	G
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G
Neurotrophic tyrosine kinase receptor 1	NTRK1	G
Neurotrophin 3	NTF3 or NT3	G
Neurturin	NRTN	G
Niacin receptor		G
Nibrin	NBS1	G
Nodal	NODAL	G
Noggin	NOG	G
Norrie disease protein	NDP	G
Notch 1	NOTCH1	G
Notch 2	NOTCH2	G
Notch 3	NOTCH3	G
Notch ligand - jagged 1	JAG1, AGS	G
Nuclear factor of activated T cells (NFAT) complex, cytosolic	NFATC	G
Nuclear factor of activated T cells (NFAT) complex, preexisting component	NFATP	G
Nuclear mitotic apparatus protein 1	NUMA1	G
Oligophrenin-1	OPHN1	G
Oncogene abl1	ABL1	G
Oncogene abl2		G
Oncogene akt1		G
Oncogene akt2	AKT2	G
Oncogene axl	AXL	G
Oncogene bcl2		G
Oncogene bcr/abl		G
Oncogene B-lym		G

Oncogene B-raf		G
Oncogene clk1		G
Oncogene c-myc		G
Oncogene cot		G
Oncogene crk		G
Oncogene crkl		G
Oncogene ect2		G
Oncogene ELK1	ELK1	G
Oncogene ELK2	ELK2	G
Oncogene ems1		G
Oncogene ERB		G
Oncogene ERB2		G
Oncogene ERBA		G
Oncogene ERBAL2		G
Oncogene ERG (early reponse gene)		G
Oncogene ETS1		G
Oncogene ETS2		G
Oncogene EVI1	EVI1	G
Oncogene fes		G
Oncogene fgr		G
Oncogene fos	FOS	G
Oncogene fps		G
Oncogene GLI1	GLI	G
Oncogene GLI2	GLI2	G
Oncogene GLI3	GLI3	G
Oncogene gro1		G
Oncogene gro2		G
Oncogene Ha-ras	HRAS	G
Oncogene hs1		G
Oncogene hst	FGF4	G
Oncogene int1	WNT1	G
Oncogene int2	FGF3	G
Oncogene int3	Notch4	G
Oncogene int4	WNT3	G
Oncogene jun	JUN	G
Oncogene KIT	KIT, PBT	G
Oncogene LCO	LCO	G
Oncogene l-myc		G
Oncogene lpsa		G
Oncogene lyn		G
Oncogene maf		G
Oncogene mas1		G
Oncogene mcf2		G
Oncogene mdm2	MDM2	G
Oncogene mel		G
Oncogene met	MET	G
Oncogene mos		G
Oncogene mpl		G
Oncogene MUM1	MUM1	G
Oncogene myb	MYB	G

Oncogene myc	MYC	G
Oncogene n-myc		G
Oncogene N-ras (neuroblastoma v-ras)	NRAS	G
Oncogene ovc		G
Oncogene pim1		G
Oncogene pti-1sea		G
Oncogene pvt1		G
Oncogene raf	RAF	G
Oncogene ralb		G
Oncogene rel		G
Oncogene ret	RET	G
Oncogene r-myc		G
Oncogene ros		G
Oncogene R-ras		G
Oncogene sis	PDGFB	G
Oncogene ski		G
Oncogene sno		G
Oncogene spi1		G
Oncogene src		G
Oncogene tc21		G
Oncogene TEL	ETV6	G
Oncogene tim		G
Oncogene vavtrk		G
Oncogene v-Ki-ras2	KRAS2	G
Oncogene yes		G
Oncogene yuasa		G
Oncostatin M	OSM	G
Oncostatin M receptor	OSMR	G
Orexin	OX	G
Orexin 1 receptor	OX1R	G
Orexin 2 receptor	OX2R	G
Orthodenticle (Drosophila) homolog 1	OTX1	G
Orthodenticle (Drosophila) homolog 2	OTX2	G
Osteonectin	ON	G
Osteopontin	OPN	G
Osteoprotegerin	OPG	G
p21-activated kinase 3	PAK3	G
Paired box homeotic gene 1	PAX1	G
Paired box homeotic gene 2	PAX2	G
Paired box homeotic gene 3	PAX3	G
Paired box homeotic gene 6	PAX6	G
Paired box homeotic gene 7	PAX7	G
Paired box homeotic gene 8	PAX8	G
Paired-like homeodomain transcription factor 2	PITX2	G
Paired-like homeodomain transcription factor 3	PITX3	G
Parathyroid hormone	PTH	G
Parathyroid hormone receptor	PTHR1	G
Parathyroid hormone related-peptide	PTHrP	G
Parathyroid hormone-like hormone	PTHLH	G
Parvalbumin	PVALB	G

Patched (Drosophila) homolog, PTCH	PTCH	G
Phosphatase & tensin homolog	PTEN	G
Phosphate regulating gene with homologies to endopeptidases on the X chromosome	PHEX	G
Phosphatidylinositol glycan, class A (paroxysmal nocturnal hemoglobinuria)	PIGA	G
Phosphatidylinositol transfer protein	PITPN	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 1	PDNP1	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 2	PDNP2	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 3	PDNP3	G
Phosphomannomutase 1	PMM1	G
Phosphomannomutase 2	PMM2	G
Phytanoyl-CoA hydroxylase	PHYH	G
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Poly(A) binding protein 2	PABP2	G
POU domain, class 1, transcription factor 1 (Pit1)	POU1F1	G
POU domain, class 3, transcription factor 4	POU3F4	G
POU domain, class 4, transcription factor 3	POU4F3	G
Pre-B-cell leukemia transcription factor 1	PBX1	G
Preproglucagon	GCG;GLP1; GLP2	G
Profibrinolysin		G
Progesterone receptor (RU486 binding receptor)	PGR	G
Prohibitin	PHB	G
Prolactin	PRL	G
Prolactin receptor	PRLR	G
Prolactin releasing hormone	PRH	G
Proliferin	PLF	G
Pro-melanin-concentrating hormone	PMCH	G
Promyelocytic leukemia gene	PML	G
Prophet of Pit1	PROP1	G
Prostaglandin (PG) D synthase, hematopoietic	PGDS	E
Prostaglandin isomerase		G
Prostaglandin-endoperoxidase synthase 2	PTGS2	G
Prostate cancer anti-metastasis gene KAI1	KAI1	G
Protein tyrosine phosphatase, non-receptor type 12	PTPN12	G
RAD51, DNA repair protein	RAD51	G
RAD52, DNA repair protein	RAD52	G
RAD54, DNA repair protein	RAD54	G
RAD55, DNA repair protein	RAD55	G
RAD57, DNA repair protein	RAD57	G
Ras-G-protein	RAS	G
Rathke pouch homeobox, RPX	RPX	G
Receptor tyrosine kinase (RTK), Nsk2	NSK2	G
Recombination activating gene 1	RAG1	G
Recombination activating gene 2	RAG2	G
Relaxin H1	RLN1	G
Relaxin H2	RLN2	G
Retinoblastoma 1	RB1	G
Retinoic acid receptor, alpha	RARA	G

Retinoic acid receptor, beta	RARB	G
Retinoic acid receptor, gamma	RARG	G
Retinoid X receptor, alpha	RXRA	G
Retinoid X receptor, beta	RXRB	G
Retinoid X receptor, gamma	RXRG	G
Retinoschisis, X-linked, juvenile	RS	G
Rhabdoid tumors	SMARCB1	G
RIGUI	RIGUI	G
Ryanodine receptor 1, skeletal	RYR1	G
SA homolog	SAH	G
Sal-like 1	SALL1	G
Serine/threonine kinase 11	STK11	G
Serine/threonine kinase 2	STK2	G
Sex determining region Y, SRY	SRY	G
Short stature homeobox	SHOX	G
Sialoprotein, bone	BSP	G
Signal transducer and activator of transcription 1	STAT1	G
Signal transducer and activator of transcription 2	STAT2	G
Signal transducer and activator of transcription 3	STAT3	G
Signal transducer and activator of transcription 4	STAT4	G
Signal transducer and activator of transcription 5	STAT5	G
Sine oculis homeobox, drosophila, homolog 1	SIX1	G
Sine oculis homeobox, drosophila, homolog 2	SIX2	G
Sine oculis homeobox, drosophila, homolog 5	SIX5	G
Slug protein		G
Smoothelin	SMTN	G
Smoothened (Drosophila) homolog	SMOH	G
Somatotrophin		G
Sonic hedgehog, SHH	SHH	G
SOS1 guanine nucleotide exchange factor	SOS1	G
Spastic paraplegia 7	SPG7	G
Sperm adhesion molecule	SPAM1	G
Sperm protamine P1	PRM1	G
Sperm protamine P2	PRM2	G
Split hand/foot malformation gene	DSS1	G
SRY-box 10	SOX10	G
SRY-box 11	SOX11	G
SRY-box 3	SOX3	G
SRY-box 4	SOX4	G
SRY-box 9	SOX9	G
Stem cell factor	SCF	G
Steroid hormone receptor responsive DNA elements		G
Stromal derived factor 1	SDF1	G
Sulfamidase	SGSH	G
Sulfonylurea receptor	SUR	G
Suppression of tumorigenicity 3 gene	ST3	G
Suppression of tumorigenicity 8 gene	ST8	G
Surfeit 1	SURF1	G
Syndecan 1	SYND1	G
Syndecan 2	SYND2	G

Syndecan 3	SYND3	G
Syndecan 4	SYND4	G
Synovial sarcoma gene 1	SSX1	G
Synovial sarcoma gene 2	SSX2	G
Talin	TLN	G
TATA binding protein	TBP	G
TATA binding protein associated factor 2A	TAF2A	G
TATA binding protein associated factor 2C2	TAF2C2	G
TATA binding protein associated factor 2D	TAF2E	G
TATA binding protein associated factor 2F	TAF2F	G
TATA binding protein associated factor 2H	TAF2H	G
TATA binding protein associated factor 2I	TAF2I	G
TATA binding protein associated factor 2J	TAF2J	G
TATA binding protein associated factor 2K	TAF2K	G
T-BOX 1	TBX1	G
T-BOX 2	TBX2	G
T-BOX 3	TBX3	G
T-BOX 4	TBX4	G
T-BOX 5	TBX5	G
T-BOX 6	TBX6	G
Testis-specific protein Y	TSPY	G
Thrombopoietin	THPO	G
Thrombospondin	THBS1	G
Thymopoietin	TMPO	G
Thyroglobulin	TG	G
Thyroid hormone receptor, alpha	THRA	G
Thyroid hormone receptor, beta	THRB	G
Thyroid peroxidase	TPO	G
Thyroid receptor auxiliary protein	TRAP	G
Thyroid-stimulating hormone receptor	TSHR	G
Thyroid-stimulating hormone, alpha	TSHA	G
Thyroid-stimulating hormone, beta	TSHB	G
Thyrotroph embryonic factor	TEF	G
Thyrotropin releasing hormone	TRH	G
Thyrotropin releasing hormone receptor	TRHR	G
TIE receptor tyrosine kinase	TIE-1	G
Torticollis, keloids, cryptorchidism and renal dysplasia gene	TKCR	G
Transcription factor 1, hepatic	TCF1	G
Transcription factor 2, hepatic	TCF2	G
Transcription factor 3	TCF3	G
Transcription factor binding to IGHM enhancer 3	TFE3	G
Transcription termination factor, RNA polymerase 1	TTF1	G
Transcription termination factor, RNA polymerase 2	TTF2	G
Transcription termination factor, RNA polymerase 3	TTF3	G
Transferrin	TF	G
Transferrin receptor	TFRC	G

Transforming growth factor, alpha	TGFA	G
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta induced	TGFBI	G
Transforming growth factor, beta receptor 2	TGFBR2	G
Transglutaminase 1	TGM1	G
Transglutaminase 2	TGM2	G
Transglutaminase 4	TGM4	G
Translocation in renal carcinoma on chromosome 8 gene	TRC8	G
Treacle gene	TCOF1	G
Tubby-like protein 1	TULP1	G
Tuberous sclerosis 1	TSC1	G
Tuberous sclerosis 2	TSC2	G
Tumor susceptibility gene 101	TSG101	G
Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tumour protein p73	TP73	G
Tumour protein, translationally-controlled 1	TPT1	G
Twist (Drosophila) homolog	TWIST	G
Ubiquitin		G
Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
Ubiquitin carboxyl-terminal esterase L1	UCHL1	G
Ubiquitin fusion degeneration 1-like	UFD1L	G
Vascular endothelial growth factor	VEGF	G
Vasoinhibitory peptide		G
Vitamin B12-binding (R) protein		G
Vitamin D receptor	VDR	G
v-myc avian myelocytomatosis viral oncogene homolog	MYC	G
Von Hippel-Lindau gene	VHL	G
Werner syndrome helicase	WRN	G
Wilms tumour gene 1	WT1	G
Wilms tumour gene 2	WT2	G
Wilms tumour gene 4	WT4	G
Winged helix nude	WHN	G
Wingless family, wnt2	WNT2	G
Wingless family, wnt4	WNT4	G
Wingless family, wnt5	WNT5	G
Wingless family, wnt7	WNT7	G
Wingless family, wnt8	WNT8	G
Wnt inhibitory factor, WIF-1	WIF1	G
Wolf-Hirschhorn syndrome candidate 1 gene	WHSC1	G
X (inactive)-specific transcript	XIST	G
X-ray repair gene	XRCC9	G
YY1 transcription factor	YY1	G
Zona pellucida glycoprotein 1	ZP1	G
Zona pellucida glycoprotein 2	ZP2	G
Zona pellucida glycoprotein 3	ZP3	G
Zona pellucida receptor tyrosine kinase	ZRK	G

Zonadhesin ZAN G

The core list of genes provides a platform for the design and application of profiling technologies to healthcare management. We have termed these designs for profiling "GenosticsTM" - an amalgam of genomics and prognosis.

This "GenosticTM" profiling of patients and persons will radically enhance the ability of clinicians, healthcare professionals and other parties to plan and manage healthcare provision and the targeting of appropriate healthcare resources to those deemed most in need.

The use of our invention could also lead to a host of new applications for such profiling technologies, such as identification of persons with particular work or environment related risk, selection of applicants for employment, training or specific opportunities or for the enhancing the planning and organisation of health services, education services and social services.

REFERENCES

Brenman, J. E.; Chao, D. S.; Xia, H.; Aldape, K.; Bredt, D. S.
Nitric oxide synthetase complexed with dystrophin and absent from skeletal muscle sarcolemma in Duchenne muscular dystrophy. Cell 82: 743-752, 1995.

British National Formulary Number 35. British Medical Association and Royal Pharmaceutical Society of Great Britain (March 1998).

Brody T.M., Larner J., Minneman K.P. Human Pharmacology Molecular to Clinical. 3rd Ed. Mosby, 1998.

Buetow KH, Edmonson MN and Cassidy AB. Reliable identification of large numbers of candidate SNP's from public EST data. Nature Genetics 21, 323- 5, 1999.

Cowen, S.G., Pipeline pulse, March 1999

Crooke ST. 1998. Optimising the impact of genomics on drug discovery and development. Nature Biotechnology 16 (Supplement): 29-30.

Davies, D. R.; Armstrong, J. G.; Thakker, N.; Horner, K.; Guy, S. P.; Clancy, T.; Sloan, P.; Blair, V.; Dodd, C.; Warnes, T. W.; Harris, R.; Evans, D. G. R.:
Severe Gardner syndrome in families with mutations restricted to a specific region of the APC gene. *Am. J. Hum. Genet.* 57: 1151-1158, 1995.

Dental Practitioners' Formulary, 1998-2000 Edition. British Medical Association and Royal Pharmaceutical Society of Great Britain (1998).

Deuter, R.; Muller, O.:Detection of APC mutations in stool DNA of patients with colorectal cancer by HD-PCR. *Hum. Mutat.* 11: 84-89, 1998.
Drew S (1998). HRT – More good than harm. Geriatric Medicine 28: 23-26.

Drmanac S, Kita D, Labat I, Hauser B, Schmidt C, Burczak JD and Drmanac R. 1998. Accurate sequencing by hybridisation for DNA diagnostics and individual genomics. Nature Biotechnology 16:54-58.

Drews. J. 1997. Nature Biotechnology14: 1516-1518.

Drews J and Ryser S. 1997. Nature Biotechnology 15: 1318-1319.

Fogarty T.C. and Ireson N.S. (1994) Evolving Bayseian classifiers for credit control – a comparison with other machine –learning methods. IMA Journal of Mathematics Applied in Business and Industry 5, 63-75.

Fox S 1998. Genomic Diagnostics: the next stage in drug development.

Gilles PN. 1999. Single nucleotide polymorphic discrimination by an electronic dot blot assay on semiconductor microchips. Nature Biotechnology 17 (4): 365-370.

Goldberg D.E. (1989) Genetic algorithms in search optimisation and machine learning. Addison-Wesley.

Goodman and Gillman. The Pharmacological Basis of Therapeutics, 9th Ed. McGraw-Hill, New York 1996.

Griffin TJ., Tang W and Smith LM. 1997. Genetic analysis by peptide nucleic acid affinity MALDI-TOF mass spectrometry. Nature Biotechnology 15 1368-1372.

Herbal Medicines, 1998

Lander ES. 1996. The new genomics: global views of biology. Science 274:540-545.

Lazarou J, Pomeranz BH, Corey PN. 1998. Incidence of adverse drug reactions in hospitalised patients: a meta-analysis of prospective studies. JAMA Apr 15; 279 (15): 1200-5

Marshall A. 1997. Laying the foundations for personalised medicines. Nature Biotechnology. 15: 954-956.

Marshall. 1997. Getting the right drug into the right patient. Nature Biotechnology. 151249-1252.

Magee, T.; Fuentes, A. M.; Garban, H.; Rajavashisth, T.; Marquez, D.; Rodriguez, J. A.; Rajfer, J.; Gonzalez-Cadavid, N. F. Cloning of a novel neuronal nitric oxide synthase expressed in penis and lower urinary tractBiochem. Biophys. Res. Commun226: 145-151, 1996.

Maher, E. R.; Barton, D. E.; Slatter, R.; Koch, D. J.; Jones, M. H.; Nagase, H.; Payne, S. J.; Charles, S. J.; Moore, A. T.; Nakamura, Y.; Ferguson-Smith, M. A.: Evaluation of molecular genetic diagnosis in the management of familial adenomatous polyposis coli: a population based study. *J. Med. Genet.* 30: 675-678, 1993.

Marshall A and Hodgson J 1998. DNA chips: an array of possibilities. Nature Biotechnology 16:27-31.

Martindale, 1998. Royal Pharmaceutical Society of Great Britain.

Nelson, R. J.; Demas, G. E.; Huang, P. L.; Fishman, M. C.; Dawson, V. L.; Dawson, T. M.; Snyder, S. H. : Behavioural abnormalities in male mice lacking neuronal nitric oxide synthase. Nature 378: 383-386, 1995.

Nickerson DA, et al DNA sequence diversity in a 9.7 -kb region of the human lipoprotein lipase gene. Nature Genetics 19, 233-40, 1998.

Pathare SR and Paton C. ABC of mental health – psychotropic drug treatment. British Medical Journal 315: 661-664, 1997.

Petersen, G. M.; Francomano, C.; Kinzler, K.; Nakamura, Y.:
Presymptomatic direct detection of adenomatous polyposis coli (APC) gene mutations in familial adenomatous polyposis. *Hum. Genet.* 91: 307-311, 1993.

Petersen, G. M.; Shohat, T.; Brown, J.; Nakamura, Y.:
Genetic counseling for familial adenomatous polyposis (FAP) with chromosome 5q linkage information. (Abstract) *Am. J. Hum. Genet.* 45 (suppl.): A125 only, 1989.
Poste G. 1998. Molecular medicine and information-based targetted of healthcare. Nature Biotechnology. 16 (Supplement): 19-21.

Rieder MJ, Taylor SL, Clark AG and Nickerson). Sequence variation in the human angiotensin converting enzyme. Nature Genetics 22, 62 - 9, 1999.
Schafer AJ and Hawkins JR 1997. DNA variation and the future of human genetics. Nature Biotechnology 16: 33-44.

Taylor D. and Kerwin R. (1997). Clozapine's role in the treatment of schizophrenia. Prescriber - Suplement.

Tyagi s, Bratu DP and Kramer FR 1998. Multicolour molecular beacons for allele discrimination. Nature Biotechnology 16: 49-53.

U.S. Pharmacopeia, 1998.

Wang DG, et al. Large-scale identification, mapping and genotyping of single nucleotide polymorphisms in the human genome. Science 265, 2049-54, 1998.

Weatherall Dj, Ledingham JGG and Warrel DA. Eds Oxford Textbook of Medicine 3rd Edition. Oxford Medical Publications 1996.

Xie, J.; Roddy, P.; Rife, T. K.; Murad, F.; Young, A. P.: Two closely linked but separable promoters for human neuronal nitric oxide synthase gene transcriptionProc. Nat. Acad. Sci 92: 1242-1246, 1995.

CLAIMS

1. A set of nucleotide probes for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; said probes being complementary to DNA and RNA sequences of said group of genes; characterised in that said group is a core group of genes consisting of substantially all of the following:

KEY TO 'PROTEIN FUNCTION' COLUMN

E ENZYME
T TRANSPORT & STORAGE
S STRUCTURAL
I IMMUNITY
N NERVOUS TRANSMISSION
G GROWTH & DIFFERENTIATION

CORE GENE LIST	HUGO GENE SYMBOL	PROTEIN FUNCTION
11beta hydroxysteroid dehydrogenase 2	HSD11B2	E
17beta hydroxysteroid dehydrogenase 1	HSD17B1	E
17beta hydroxysteroid dehydrogenase 3	HSD17B3	E
17beta hydroxysteroid dehydrogenase 4	HSD17B4	E
17beta hydroxysteroid oxidoreductase		E
18-hydroxysteroid oxidoreductase		E
2,3-bisphosphoglycerate mutase	BPGM	E
2,4-dienoyl CoA reductase	DECR	E
3 beta hydroxysteroid dehydrogenase 2	HSD3B2	E
3-oxoacid CoA transferase	OXCT	E
4-hydroxyphenylpyruvate dioxygenase	HPD	E
5,10-methylenetetrahydrofolate reductase (NADPH)	MTHFR	E
5-adenosyl homocysteine hydrolase		E
6-phosphofructo-2-kinase	PFKFB1	E
6-pyruvoyltetrahydropterin synthase	PTS	E
Acetoacetyl 1-CoA-thiolase	ACAT1	E
Acetoacetyl 2-CoA-thiolase	ACAT2	E
Acetyl CoA acyltransferase	ACAA	E
Acetyl CoA carboxylase	ACC	E
Acetyl CoA carboxylase alpha	ACACA	E
Acetyl CoA synthase		E
Acetylcholinesterase	ACHE	E
Acid phosphatase 2, lysosomal	ACP2	E
Aconitase		E
Acyl CoA dehydrogenase, long chain	ACADL	E
Acyl CoA dehydrogenase, medium chain	ACADM	E
Acyl CoA dehydrogenase, short chain	ACADS	E
Acyl CoA dehydrogenase, very long chain	ACADVL	E
Acyl CoA synthetase, long chain, 1	LACS1	E

Acyl CoA synthetase, long chain, 2	LACS2	E
Acyl CoA synthetase, long chain, 4	ACS4	E
Acyl malonyl condensing enzyme		E
Acyl-CoA thioesterase		E
ADAM (A disintegrin and metalloproteinase) 1	ADAM1	E
ADAM (A disintegrin and metalloproteinase) 10	ADAM10	E
ADAM (A disintegrin and metalloproteinase) 11	ADAM11	E
ADAM (A disintegrin and metalloproteinase) 12	ADAM12	E
ADAM (A disintegrin and metalloproteinase) 13	ADAM13	E
ADAM (A disintegrin and metalloproteinase) 14	ADAM14	E
ADAM (A disintegrin and metalloproteinase) 15	ADAM15	E
ADAM (A disintegrin and metalloproteinase) 16	ADAM16	E
ADAM (A disintegrin and metalloproteinase) 17	ADAM17	E
ADAM (A disintegrin and metalloproteinase) 18	ADAM18	E
ADAM (A disintegrin and metalloproteinase) 19	ADAM19	E
ADAM (A disintegrin and metalloproteinase) 2	ADAM2	E
ADAM (A disintegrin and metalloproteinase) 3A	ADAM3A	E
ADAM (A disintegrin and metalloproteinase) 3B	ADAM3B	E
ADAM (A disintegrin and metalloproteinase) 4	ADAM4	E
ADAM (A disintegrin and metalloproteinase) 5	ADAM5	E
ADAM (A disintegrin and metalloproteinase) 6	ADAM6	E
ADAM (A disintegrin and metalloproteinase) 7	ADAM7	E
ADAM (A disintegrin and metalloproteinase) 8	ADAM8	E
ADAM (A disintegrin and metalloproteinase) 9	ADAM9	E
Adenosine deaminase	ADA	E
Adenosine monophosphate deaminase	AMPD	E
Adenylate cyclase 1	ADCY1	E
Adenylate cyclase 2	ADCY2	E
Adenylate cyclase 3	ADCY3	E
Adenylate cyclase 4	ADCY4	E
Adenylate cyclase 5	ADCY5	E
Adenylate cyclase 6	ADCY6	E
Adenylate cyclase 7	ADCY7	E
Adenylate cyclase 8	ADCY8	E
Adenylate cyclase 9	ADCY9	E
Adenylate kinase	AK1	E
Adenylate transferase		E
Adenylosuccinate lyase	ADSL	E
ADP-ribosyltransferase	ADPRT	E
Adrenoleukodystrophy gene	ALD	E
Alanine-glyoxylate aminotransferase	AGXT	E
Alcohol dehydrogenase 1	ADH1	E
Alcohol dehydrogenase 2	ADH2	E
Alcohol dehydrogenase 3	ADH3	E
Alcohol dehydrogenase 4	ADH4	E
Alcohol dehydrogenase 5	ADH5	E
Alcohol dehydrogenase 6	ADH6	E
Alcohol dehydrogenase 7	ADH7	E
Aldehyde dehydrogenase 1	ALDH1	E
Aldehyde dehydrogenase 10	ALDH10	E

Aldehyde dehydrogenase 2	ALDH2	E
Aldehyde dehydrogenase 5	ALDH5	E
Aldehyde dehydrogenase 6	ALDH6	E
Aldehyde dehydrogenase 7	ALDH7	E
Aldolase A	ALDOA	E
Aldolase B	ALDOB	E
Aldolase C	ALDOC	E
Alkylglycerone phosphate synthase	AGPS	E
alpha1-antichymotrypsin	AACT	E
alpha1-antitrypsin	PI	E
alpha2-antiplasmin	PLI	E
alpha-amino adipic semialdehyde synthase		E
alpha-amylase		E
alpha-dextrinase		E
alpha-Galactosidase A	GLA	E
Alpha-galactosidase B, GALB	NAGA	E
alpha-glucosidase, neutral C	GANC	E
alpha-glucosidase, neutral AB	GANAB	E
Peptidylglycine alpha-amidating monooxygenase	PAM	E
alpha-ketoglutarate dehydrogenase		E
alpha-L-Iduronidase	IDUA	E
Aminomethyltransferase	AMT	E
Aminopeptidase P	XPNPEP2	E
Amylo-1,6-glucosidase	AGL	E
Angiotensin converting enzyme	ACE, DCP1	E
Angiotensinogen	AGT	E
Antithrombin III	AT3	E
Apurinic endonuclease	APE	E
Arginase	ARG1	E
Arginosuccinate lyase	ASL	E
Arginosuccinate synthetase	ASS	E
Arylsulfatase A	ARSA	E
Arylsulfatase B	ARSB	E
Arylsulfatase C	ARSC1	E
Arylsulfatase D	ARSD	E
Arylsulfatase E	ARSE	E
Arylsulfatase F	ARSF	E
Asparagine synthetase	AS	E
Aspartate transcarbamoylase		E
Aspartoacylase	ASPA	E
Aspartylglucosaminidase	AGA	E
ATP cobalamin adenoxyltransferase		E
ATP sulphurylase	atpsk2	E
ATP/ADP translocase		E
beta-galactosidase	GLB1	E
beta-glucosidase, neutral		E
beta-Glucuronidase	GUSB	E
beta-ketoacyl reductase		E
beta-N-acetylhexosaminidase, A		E
beta-N-acetylhexosaminidase, B		E

Bile acid coenzyme A: amino acid N-acyltransferase	BAAT	E
Bile salt-stimulated lipase	CEL	E
Bilirubin UDP-glucuronosyltransferase		E
Biotinidase	BTD	E
Bleomycin hydrolase	BLMH	E
Branched chain aminotransferase 1, cytosolic	BCAT1	E
Branched chain aminotransferase 2, mitochondrial	BCAT2	E
Branched chain keto acid dehydrogenase E1, alpha polypeptide	BCKDHA	E
Branched chain keto acid dehydrogenase E1, beta polypeptide	BCKDHB	E
Brush border guanylyl cyclase		E
Butyrylcholinesterase	BCHE	E
C1 inhibitor		E
C17-20 desmolase		E
C3 convertase		E
Calpain	CAPN, CAPN3	E
Carbamoylphosphate synthetase 1	CPS1	E
Carbamoylphosphate synthetase 2	CPS2	E
Carbonic anhydrase, alpha	CA1	E
Carbonic anhydrase, beta	CA2	E
Carbonic anhydrase 3	CA3	E
Carbonic anhydrase 4	CA4	E
Carboxylesterase 1	CES1	E
Carboxypeptidase	CPN	E
Carnitine acetyltransferase	CRAT	E
Carnitine acylcarnitine translocase	CACT	E
Carnitine palmitoyltransferase I	CPT1A	E
Carnitine palmitoyltransferase II	CPT2	E
Catechol-O-methyltransferase	COMT	E
Cathepsin B		E
Cathepsin D		E
Cathepsin E		E
Cathepsin G	CTSG	E
Cathepsin H		E
Cathepsin K	CTSK	E
Cathepsin L		E
Cathepsin S		E
Caveolin 3	CAV3	E
Ceruloplasmin precursor	CP	E
Chitotriosidase	chit	E
Cholesterol ester hydroxylase		E
Choline acetyltransferase	CHAT	E
Chymase	CHY1	
Chymotrypsinogen		E
Citrate synthase		E
CoA transferase		E
Coenzyme Q (CoQ)/ubiquinone		E
Collagenic-like tail subunit of asymmetric	COLQ	E

acetylcholinesterase		
Complex I		E
Complex II		E
Complex III		E
Complex III		E
Complex V	MTATP6	E
Coproporphyrinogen oxidase	CPO	E
Creatine kinase – B and m	CKBE	E
Cu2+ transporting ATPase alpha polypeptide	ATP7A	E
Cu2+ transporting ATPase beta polypeptide	ATP7B	E
Cyclic nucleotide phosphodiesterase 1B	PDE1B	E
Cyclic nucleotide phosphodiesterase 1B1	PDE1B1	E
Cyclic nucleotide phosphodiesterase 2A3	PDE2A3	E
Cyclic nucleotide phosphodiesterase 3A	PDE3A	E
Cyclic nucleotide phosphodiesterase 3B	PDE3B	E
Cyclic nucleotide phosphodiesterase 4A	PDE4A	E
Cyclic nucleotide phosphodiesterase 4C	PDE4C	E
Cyclic nucleotide phosphodiesterase 5A	PDE5A	E
Cyclic nucleotide phosphodiesterase 6A	PDE6A	E
Cyclic nucleotide phosphodiesterase 6B	PDE6B	E
Cyclic nucleotide phosphodiesterase 7	PDE7	E
Cyclic nucleotide phosphodiesterase 8	PDE8	E
Cyclic nucleotide phosphodiesterase 9A	PDE9A	E
Cyclooxygenase 1	COX1	E
Cyclooxygenase 2	COX2	E
CYP11A1	CYP11A1	E
CYP11B1	CYP11B1	E
CYP11B2	CYP11B2	E
CYP17	CYP17	E
CYP19	CYP19	E
CYP1A1	CYP1A1	E
CYP1A2	CYP1A2	E
CYP1B1	CYP1B1	E
CYP21	CYP21	E
CYP24	CYP24	E
CYP27	CYP27	E
CYP27B1	PDDR	E
CYP2A1	CYP2A1	E
CYP2A13	CYP2A13	E
CYP2A3	CYP2A3	E
CYP2A6V2	CYP2A6V2	E
CYP2A7	CYP2A7	E
CYP2B6	CYP2B6	E
CYP2C18	CYP2C18	E
CYP2C19	CYP2C19	E
CYP2C8	CYP2C8	E
CYP2C9	CYP2C9	E
CYP2D6	CYP2D6	E
CYP2E1	CYP2E1	E
CYP2F1	CYP2F1	E

CYP2J2	CYP2J2	E
CYP3A3	CYP3A3	E
CYP3A4	CYP3A4	E
CYP3A5	CYP3A5	E
CYP3A7	CYP3A7	E
CYP4A11	CYP4A11	E
CYP4B1	CYP4B1	E
CYP4F2	CYP4F2	E
CYP4F3	CYP4F3	E
CYP51	CYP51	E
CYP5A1	CYP5A1	E
CYP7A	CYP7A	E
CYP8	CYP8	E
Cystathionase	CTH	E
Cystathione beta synthase	CBS	E
Cytidine deaminase	CDA	E
Cytidine-5-prime-triphosphate synthetase	CTPS	E
Cytochrome a		E
Cytochrome b-245 alpha	CYBA	E
Cytochrome b-245 beta	CYBB	E
Cytochrome b-5	CYB5	E
Cytochrome c		E
Cytochrome c oxidase, MTCO		E
D-beta-hydroxybutyrate dehydrogenase		E
Dehydratase		E
Delta 4-5 alpha-reductase		E
Delta 4-5 oxosteroid isomerase		E
Delta aminolevulinate dehydratase	ALAD	E
Delta aminolevulinate synthase 1	ALAS1	E
Delta aminolevulinate synthase 2	ALAS2	E
Delta(4)-3-oxosteroid 5-beta-reductase		E
Delta-7-dehydrocholesterol reductase	DHCR7	E
Deoxycorticosterone (DOC) receptor		E
Deoxycytidine kinase DCK		E
Deoxyuridine triphosphatase; dUTPase		E
DHEA sulfotransferase	STD	E
Dihydrodiol dehydrogenase 1	DDH1	E
Dihydrofolate reductase	DHFR	E
Dihydrolipoyl dehydrogenase		E
Dihydrolipoyl dehydrogenase 2	PDHA	E
Dihydrolipoyl succinyltransferase	DLST	E
Dihydrolipoyl transacetylase	PDHA	E
Dihydroorotase		E
Dihydropyramidinase	DPYS	E
Dihydroxyacetonephosphate acyltransferase	DHAPAT	E
Dihyropyrimidine dehydrogenase	DPYD	E
DM-Kinase	DMPK	E
DNA directed polymerase, alpha	POLA	E
DNA glycosylases		E
DNA helicases		E

DNA Ligase 1	LIG1	E
DNA methyltransferase	DNMT	E
Methylguanine-DNA methyltransferase	MGMT	E
DNA polymerase 1		E
DNA polymerase 2		E
DNA polymerase 3		E
DNA primase		E
DNA-dependant RNA polymerase		E
DOPA decarboxylase	DDC	E
Dopamine beta hydroxylase	DBH	E
Dysferlin	DYS, DYSF	E
Dystrophia myotonica	DM, DMPK	E
Dystrophia myotonica, atypical	DM2	E
Elastase 1	ELAS1	E
Elastase 2	ELAS2	E
Electron-transferring flavoprotein dehydrogenase	ETFDH	E
Enolase	ENO1	E
Enoyl CoA hydratase		E
Enoyl CoA isomerase		E
Enoyl CoA reductase		E
Enterokinase	PRSS7, ENTK	E
Eosinophil peroxidase	EPX	E
Epilepsy, benign neonatal 4 gene	ICCA	E
Epilepsy, female restricted	EFMR	E
Epilepsy, progressive myoclonic 2 gene	EPM2A	E
Epoxide hydrolase 1, microsomal	EPHX1	E
Excision repair complementation group 1 protein	ERCC1	E
Excision repair complementation group 2 protein	ERCC2	E
Excision repair complementation group 2 protein	ERCC3	E
Excision repair complementation group 4 protein	ERCC4	E
Excision repair complementation group 6 protein	ERCC6	E
FADH dehydrogenase		E
Ferrochelatase	FECH	E
Flavin-containing monooxygenase 1	FMO1	E
Flavin-containing monooxygenase 2	FMO2	E
Flavin-containing monooxygenase 3	FMO3	E
Flavin-containing monooxygenase 4	FMO4	E
Formiminotransferase		E
Fructose-1,6-diphosphatase	FBP1	E
Fucosidase alpha-L-1	FUCA1	E
Fucosidase alpha-L-2		E
Fumarase	FH	E
Fumarylacetoacetase	FAH	E
GABA transaminase	ABAT	E
Gadd45 (growth arrest & DNA-damage-inducible protein)		E
Galactocerebrosidase	GALC	E
Galactokinase	GALK1	E
Galactose 1-phosphate uridyl-transferase	GALT	E
Gastric Intrinsic factor, GIF	GIF	E
Glucokinase	GCK	E

Glucosaminyl (N-acetyl) transferase 2, I-branching enzyme	GCNT2	E
Glucose-6-phosphatase	G6PC	E
Glucose-6-phosphatase translocase	G6PT1	E
Glucose-6-phosphate dehydrogenase	G6PD	E
Glucosidase, acid alpha	GAA	E
Glucosidase, acid beta	GBA	E
Glutamate decarboxylase, GAD	GAD1	E
Glutamate dehydrogenase	GLUD1	E
Glutamate-cysteine ligase	GLCLC	E
Glutamine phosphoribosylpyrophosphate amidotransferase/PRPP amidotransferase		E
Glutamine synthase		E
Glutaryl-CoA dehydrogenase	GCDH	E
Glutathione peroxidase, GPX1	GPX1	E
Glutathione peroxidase, GPX2	GPX2	E
Glutathione reductase, GSR	GSR	E
Glutathione S-transferase mu 1, GSTM1	GSTM1	E
Glutathione S-transferase mu 4, GSTM4		E
Glutathione S-transferase theta 1, GSTT1	GSTT1	E
Glutathione S-transferase theta 2, GSTT2		E
Glutathione S-transferase, GSTP1	GSTP1	E
Glutathione S-transferase, GSTZ1	GSTZ1	E
Glutathione synthetase	GSS	E
Glyceraldehyde-3-phosphate dehydrogenase, GAPDH	GAPDH	E
Glycerol kinase	GK	E
Glycerophosphate dehydrogenase 2	GPD2	E
Glycinamide ribonucleotide (GAR) transformylase	GART	E
Glycine dehydrogenase	GLDC	E
Glycogen branching enzyme	GBE1	E
Glycogen phosphorylase	PYGL	E
Glycogen synthase 1 (muscle)	GLYS1	E
Glycogen synthase 2 (liver)	GYS2	E
Glycosyltransferases, ABO blood group	ABO	E
GM2 ganglioside activator protein, GM2A	GM2A	E
Guanidinoacetate N-methyltransferase	GAMT	E
Guanylate cyclase 2D, membrane (retina-specific)	GUCY2D	E
Guanylate cyclase activator 1A (retina)	GUCA1A	E
Guanylate kinase		E
Guanylyl cyclase		E
Haeme regulated inhibitor kinase		E
Heparan sulfamidase		E
Hepatic lipase	LIPC	E
Hepatic nuclear factor-3-beta	HNF3B	E
Hepatic nuclear factor-4-alpha	HNF4A	E
Hexokinase 1	HK1	E
Hexokinase 2	HK2	E
Hexosaminidase A	HEXA,TSD	E
Hexosaminidase B	HEXB	E

Histidase		E
HMG-CoA lyase	HMGCL	E
HMG-CoA reductase	HMGCR	E
HMG-CoA synthase	HMGCS2	E
Holocarboxylase synthetase	HLCS	E
Homogentisate 1,2 dioxygenase	HGD	E
Hormone-sensitive lipase	HSL	E
HSSB, replication protein		E
Hydroxyacyl glutathione hydrolase	HAGH	E
Hypoxanthine-guanine phosphoribosyltransferase, HGPRT	HPRT	E
Hypoxia inducible factor 1	HIF1A	E
Hypoxia inducible factor 2		E
Ibonucleoside diphosphate reductase		E
Iduronate 2 sulphatase	IDS	E
Inosine monophosphate dehydrogenase, IMPDH		E
Inosine triphosphatase	ITPA	E
Inter-alpha-trypsin inhibitor, IATI		E
Iodothyronine-5'-deiodinase, type 1 and 2		E
IP3 kinase		E
Isocitrate dehydrogenase		E
Isovaleric acid CoA dehydrogenase	IVD	E
Ketohexokinase	KHK	E
ketolase		E
Kynurenine hydroxylase		E
Kynureninease		E
Lactase		E
Lactate dehydrogenase, A	LDHA	E
Lactate dehydrogenase, B	LDHB	E
Lecithin-cholesterol acyltransferase	LCAT	E
Leukotriene A4 synthase	LTA4S	E
Leukotriene B4 synthase	LTB4S	E
Leukotriene C4 synthase	LTC4S	E
Lipoamide dehydrogenase	OGDH	E
Lipoxygenase		E
Lowe oculocerbrorenal syndrome gene	OCRL	E
Lysosomal acid lipase	LIPA	E
Lysyl hydroxylase	PLOD	E
Lysyl oxidase	LOX	E
Malate dehydrogenase, mitochondrial	MDH2	E
Malonyl CoA decarboxylase		E
Malonyl CoA transferase		E
Maltase-glucoamylase		E
Mannosidase, alpha B lysosomal	MANB	E
Mannosidase, beta A lysosomal	MANBA	E
Matrix metalloproteinase 1	MMP1	E
Matrix metalloproteinase 10	MMP10	E
Matrix metalloproteinase 11	MMP11	E
Matrix metalloproteinase 12	MMP12	E
Matrix metalloproteinase 13	MMP13	E

Matrix metalloproteinase 14	MMP14	E
Matrix metalloproteinase 15	MMP15	E
Matrix metalloproteinase 16	MMP16	E
Matrix metalloproteinase 17	MMP17	E
Matrix metalloproteinase 18	MMP18	E
Matrix metalloproteinase 19	MMP19	E
Matrix metalloproteinase 2	MMP2	E
Matrix metalloproteinase 3	MMP3, STMY1	E
Matrix metalloproteinase 4	MMP4	E
Matrix metalloproteinase 5	MMP5	E
Matrix metalloproteinase 6	MMP6	E
Matrix metalloproteinase 7	MMP7	E
Matrix metalloproteinase 8	MMP8	E
Matrix metalloproteinase 9	MMP9	E
MEK kinase, MEKK		E
Methionine adenosyltransferase	MAT1A, MAT2A	E
Methionine synthase	MTR	E
Methionine synthase reductase	MTRR	E
Methylmalonyl-CoA mutase	MUT	E
Mevalonate kinase	MVK	E
Mitochondrial trifunctional protein, alpha subunit	HADHA	E
Mitochondrial trifunctional protein, beta subunit	HADHB	E
Molybdenum cofactor synthesis 1	MOCS1	E
Molybdenum cofactor synthesis 2	MOCS2	E
Monoamine oxidase A	MAOA	E
Monoamine oxidase B	MAOB	E
Mucolipidoses	GNPTA	E
Muscle phosphorylase	PYGM	E
N-acetylgalactosamine-6-sulfate sulfatase	GALNS	E
N-acetylglucosamine-6-sulfatase	GNS	E
N-acetylglucosaminidase, alpha	NAGLU	E
N-acetyltransferase 1	NAT1	E
N-acetyltransferase 2	NAT2	E
NADH dehydrogenase		E
NADH dehydrogenase (ubiquinone) Fe-S protein 1	NDUFS1	E
NADH dehydrogenase (ubiquinone) Fe-S protein 4	NDUFS4	E
NADH dehydrogenase (ubiquinone) flavoprotein 1	NDUFV1	E
NADH-cytochrome b5 reductase	DIA1	E
NADPH-dependent cytochrome P450 reductase	POR	E
Neuroendocrine convertase 1	NEC1, PCSK1	E
Neutral endopeptidase		E
Nitric oxide synthase 1, NOS1	NOS1	E
Nitric oxide synthase 2, NOS2	NOS2	E
Nitric oxide synthase 3, NOS3	NOS3	E
Nucleoside diphosphate kinase-A	NDPKA	E
Ornithine delta-aminotransferase	OAT	E
Ornithine transcarbamoylase	OTC, NME1	E
Pancreatic amylase		E
Pancreatic lipase	PNLIP	E
Pancreatic lipase related protein 1	PLRP1	E

Pancreatic lipase related protein 2	PLRP2	E
Paraoxonase PON1	PON1	E
Paraoxonase PON2	PON2	E
Paraoxonase PON3		E
PCNA (proliferating cell nuclear antigen)		E
Pepsinogen		E
Peroxidase, salivary	SAPX	E
Phenylalanine hydroxylase	PAH	E
Phenylalanine monooxygenase		E
Phenylethanolamine N-methyltransferase, PNMT	PNMT	E
Phosphoenolpyruvate carboxykinase	PCK1	E
Phosphofructokinase, liver	PFKL	E
Phosphofructokinase, muscle	PFKM	E
Phosphoglucomutase		E
Phosphoglucose isomerase	GPI	E
Phosphoglycerate kinase 1	PGK1	E
Phosphoglycerate mutase 2	PGAM2	E
Phosphoribosyl pyrophosphate synthetase	PRPS1	E
Phosphorylase kinase deficiency, liver	PHK	E
Phosphorylase kinase, alpha 1 (muscle)	PHKA1	E
Phosphorylase kinase, alpha 2	PHKA2	E
Phosphorylase kinase, beta	PHKB	E
Phosphorylase kinase, delta		E
Phosphorylase kinase, gamma 2	PHKG2	E
Pineolytic beta-receptors		E
Plasminogen	PLG	E
Plasminogen activator inhibitor 1	PAI1	E
Plasminogen activator inhibitor 2	PAI2	E
Plasminogen activator receptor, Urokinase	UPAR; PLAUR	S
Plasminogen activator, Tissue	PLAT; TPA	E
Plasminogen activator, Urokinase	UPA; PLAU	E
Poly (ADP-ribose) synthetase	PARS	E
Porphobilinogen deaminase	HMBS	E
Procollagen N-protease		E
Procollagen peptidase		E
Proline dehydrogenase	PRODH	E
Prolyl-4-hydroxylase		E
Propionyl-CoA carboxylase, alpha	PCCA	E
Propionyl-CoA carboxylase, beta	PCCB	E
Prostasin, PRSS8	PRSS8	E
Protease nexin 2	PN2	E
Protective protein for beta-galactosidase	PPGB	E
Protein kinase A		E
Protein kinase B	PRKB	
Protein kinase C, alpha	PRKCA	E
Protein kinase C, gamma	PRKCG	E
Protein kinase DNA-activated	PRKDC	E
Protein kinase G		E
Protein phosphatase 1, regulatory (inhibitor) subunit 3	PPP1R3	E

Protein phosphatase 2, regulatory subunit A, beta isoform	PPP2R1B	E
Protoporphyrinogen oxidase	PPOX	E
Pterin-4-alpha-carbinolamine	PCBD	
Purine nucleoside phosphorylase	NP	E
Pyrroline-5-carboxylate synthetase	PYCS	E
Pyruvate carboxylase	PC	E
Pyruvate decarboxylase	PDHA	E
Pyruvate kinase	PKLR	E
Quinoid dihydropteridine reductase	QDPR	E
Renin	REN	E
Replication factor A		E
Replication factor C	RFC2	E
Rhodopsin kinase	RHOK	E
Ribonucleotide reductase, RRM		E
Ribosephosphate pyrophosphokinase		E
Ribosomal protein L13A	RPL13A	G
Ribosomal protein L17	RPL17	G
Ribosomal protein S19	RPS19	E
Ribosomal protein S4, X-linked	RPS4X	E
Ribosomal protein S6 kinase	RPS6KA3	E
Ribosomal protein S9	RPS9	G
S-adenosylmethionine decarboxylase, AMD		E
Serine hydroxymethyltransferase	SHMT	E
Serotonin N-acetyltransferase	SNAT	E
Sorbitol dehydrogenase	SORD	E
Sphingomyelinase	SMPD1	E
Steroid 5 alpha reductase 1	SRD5A1	E
Steroid 5 alpha reductase 2	SRD5A2	E
Steroid sulphatase	STS	E
Succinate dehydrogenase 1	SDH1	E
Succinate dehydrogenase 2	SDH2	E
Succinate thiokinase		E
Succinic semi-aldehyde dehydrogenase	ssadh	E
Succinyl CoA synthase		E
Sucrase		E
Sulfite oxidase	SUOX	E
Superoxide dismutase 1	SOD1	E
Superoxide dismutase 3	SOD3	E
TEK, tyrosine kinase, endothelial	TEK	E
Telomerase protein component		E
Terminal deoxynucleotidyltransferase, TDT		E
Thiolase, perioxisomal		E
Thiopurine S-methyltransferase	TPMT	E
Thymidylate synthase	TYMS	E
Tissue inhibitor of metalloproteinase 1, TIMP1	TIMP1	E
Tissue inhibitor of metalloproteinase 2, TIMP2	TIMP2	E
Tissue inhibitor of metalloproteinase 3, TIMP3	TIMP3	E
Tissue inhibitor of metalloproteinase 4, TIMP4	TIMP4	E
Tissue non-specific alkaline phosphatase TNSAP		E

Topoisomerase I		E
Topoisomerase II		E
Transacylase		E
Transketolase	TKT	E
Transketolase-like 1	TKTL1	E
Triosephosphate isomerase	TPI1	E
Trypsin inhibitor		E
Trypsinogen 1	TRY1	E
Trypsinogen 2	TRY2	E
Tryptophan hydroxylase	TPH	E
Tyrosinase	TYR	E
Tyrosinase-related protein 1	TYRP1	E
Tyrosine aminotransferase	TAT	E
Tyrosine hydroxylase	TH	E
Ubiquitin activating enzyme, E1		E
Ubiquitin protein ligase E3A	UBE3A	E
UDP-glucose pyrophosphorylase		E
UDP-glucuronosyltransferase 1	ugt1d, UGT1	E
UDP-glucuronosyltransferase 2	UGT2	E
Urate oxidase	UOX	E
Ureidopropionase		E
Uridinediphosphate(UDP)-galactose-4-epimerase	GALE	E
Uroporphyrinogen decarboxylase	UROD	E
Uroporphyrinogen III synthase	UROS	E
Xanthine dehydrogenase	XDH	E
Xeroderma pigmentosum, complementation group A	XPA	E
Xeroderma pigmentosum, complementation group B	XPB	E
Xeroderma pigmentosum, complementation group C	XPC	E
Xeroderma pigmentosum, complementation group D		E
Xeroderma pigmentosum, complementation group E		E
Xeroderma pigmentosum, complementation group F	XPF	E
Xeroderma pigmentosum, complementation group G	ERCC5	E
Xylitol dehydrogenase		E
Acidic amino acid transporter		T
Adaptin, beta 3A	ADTB3A	T
Adenine phosphoribosyltransferase	APRT	T
Alanine aminotransferase		T
Albumin, ALB	ALB	T
Aldose reductase		T
Alkaline phosphatase, liver/bone/kidney	ALPL	T
Alpha 1 acid glycoprotein	AAG; AGP	T
Androgen binding protein	ABP	T
Angiotensin receptor 1	AGTR1	T

Angiotensin receptor 2	AGTR2	T
Antidiuretic hormone receptor	ADHR	T
Apolipoprotein (a)	LPA	T
Apolipoprotein A 4	APOA4	T
Apolipoprotein A I	APOA1	T
Apolipoprotein A II	APOA2	T
Apolipoprotein B	APOB	T
Apolipoprotein C1	APOC1	T
Apolipoprotein C2	APOC2	T
Apolipoprotein C3	APOC3	T
Apolipoprotein D	APOD	T
Apolipoprotein E	APOE	T
Apolipoprotein H	APOH	T
Aquaporin 1	AQP1	T
Aquaporin 2	AQP2	T
Aryl hydrocarbon receptor	AHR	T
Aryl hydrocarbon receptor nuclear translocator	ARNT	T
Aspartate transaminase		T
Bestrophin	VMD2	T
Bile salt export pump	BSEP, PFIC2	T
Biliverdin reductase		T
Ca(2+) transporting ATPase, fast twitch	ATP2A1	T
Ca(2+) transporting ATPase, slow twitch	ATP2A2	T
Calcium sensing receptor	CASR	T
Calmodulin dependant kinase		T
Canalicular multispecific organic anion transporter	CMOAT	T
Carnitine transporter protein	CDSP, SCD	T
Chediak-Higashi syndrome 1 gene	CHS1	T
Cholesterol ester transfer protein	CETP	T
Clathrin		T
Cortico-steroid binding protein		T
Corticotrophin-releasing hormone	CRH	T
Corticotrophin-releasing hormone receptor	CRHR1	T
Cubilin	CUBN	T
Cystatin B	CSTB	T
Cystatin C	CST3	T
Cysteine-rich intestinal protein		T
Cystinosin	CTNS	T
Diastrophic dysplasia sulfate transporter	DTD	T
Duffy blood group	FY	T
Electron-transfering-flavoprotein alpha	ETFA	T
Electron-transfering-flavoprotein beta	ETFB	T
Emerin	EMD	T
Enteric lipase		T
Faciogenital dysplasia	FGD1, FGDY	T
Fanconi anemia, complementation group A	FANCA	T
Fanconi anemia, complementation group C	FANCC	T
Fanconi anemia, complementation group D	FANCD	T
Fatty acid binding proteins FABP1		T
Fatty acid binding proteins FABP2	FABP2	T

Fatty acid binding proteins FABP3		T
Fatty acid binding proteins FABP4		T
Fatty acid binding proteins FABP5		T
Fatty acid binding proteins FABP6		T
Ferritin, H subunit		T
Ferritin, L subunit	FTL	T
Fucosyltransferase 2	FUT2	T
Fucosyltransferase 3	FUT3	T
Fucosyltransferase 6	FUT6	T
Furin		T
Gamma-glutamyl carboxylase	GGCX	T
Gamma-glutamyltransferase 1	GGT1	T
Gamma-glutamyltransferase 2	GGT2	T
Gap junction protein alpha 1	GJA1	T
Gap junction protein alpha 3	GJA3	T
Gap junction protein alpha 8	GJA8	T
Gap junction protein beta 1	GJB1	T
Gap junction protein beta 2	GJB2	T
Gap junction protein beta 3	GJB3	T
Gastric inhibitory polypeptide GIP	GIP	T
Gastric inhibitory polypeptide receptor, GIPR	GIPR	T
Gastric lipase, LIPF		T
Gastrin releasing peptide	GRP	T
Gastrin releasing peptide receptor	GRPR	T
Glucagon synthase		T
Glutamine transporter		T
Glutathione	GSH	T
Guanylin	GUCA2	T
Haem oxygenase		T
Haemoglobin alpha 1	HBA1	T
Haemoglobin alpha 2	HBA2	T
Haemoglobin beta	HBB	T
Haemoglobin delta	HBD	T
Haemoglobin epsilon		T
Haemoglobin gamma A	HBG1	T
Haemoglobin gamma B	HBG2	T
Haemoglobin gamma G	HBGG	T
Hemochromatosis	HFE	T
Hermansky-pudlak syndrome gene	HPS	T
Histidine-rich glycoprotein	HRG	T
Huntingtin	HD	T
Hyaluronidase		T
Intestinal alkaline phosphatase IAP		T
Kell blood group precursor	XK, KEL	T
Lactotransferrin	LTF	T
Lipoprotein receptor, Low Density	LDLR	T
Lipoprotein, High Density	HDLDT1	T
Lipoprotein, Intermediate Density		T
Lipoprotein, Low Density 1		T
Lipoprotein, Low Density 2		T

Lipoprotein, Very Low Density	VLDLR	T
Long QT-type 2 potassium channels	LQT2, KCNH2	T
Low density lipoprotein receptor-related protein precursor	LRP	T
Mannosyl (alpha-1,6-)-glycoprotein beta-1, 2-N-acetylglucosaminyltransferase	MGAT2	T
Marenostrin	MEFV	T
Melanocortin 1 receptor	MC1R	T
Melanocortin 2 receptor	MC2R	T
Melanocortin 4 receptor	MC4R	T
Metallothionein		T
Microsomal triglyceride transfer protein	MTP	T
Mucin 18	MUC18	T
Mucin, MUC2		T
Mucin, MUC5AC		T
Mucin, MUC6		T
Mulibrey nanism	MUL	T
Myocilin	MYOC	T
Myoglobin		T
Myopia 1	MYP1	T
Myopia 2	MYP2	T
Na+/H+ exchanger 1	NHE1	T
Na+/H+ exchanger 2	NHE2	T
Na+/H+ exchanger 3	NHE3	T
Na+/H+ exchanger 4	NHE4	T
Na+/H+ exchanger 5	NHE5	T
Na+coupled glucose/galactose transporter		T
Nephrolithiasis 2	NPHL2	T
Nephronophthisis 1	NPHP1	T
Nephronophthisis 2	NPHP2	T
Nephrosis 1	NPHS1	T
Neuraminidase sialidase	NEU	T
Niemann-Pick disease protein	NPC1	T
Nucleophosmin	NPM1	T
Palmitoyl-protein thioesterase	PPT	T
Pancreatic colipase		T
Pendrin, PDS	PDS	T
Pepsin		T
Peptidases A		T
Peptidases B		T
Peptidases C		T
Peptidases D	PEPD	T
Peptidases E		T
Peptidases S		T
Peroxisomal membrane protein 3	PXMP3	T
Peroxisome biogenesis factor 1	PEX1	T
Peroxisome biogenesis factor 6	PEX6	T
Peroxisome biogenesis factor 7	PEX7	T
Peroxisome biogenesis factor 19	PEX19	T
Peroxisome proliferative activated receptor, alpha	PPARA	T

Peroxisome proliferative activated receptor, gamma	PPARG	T
Peroxisome receptor 1	PXR1	T
P-glycoprotein 1	PGY1	T
P-glycoprotein 3	PGY3	T
Phosphomannomutase-2	PMM2	T
Phosphomannose isomerase-1, PMI1	MPI	T
Plakophilin 1	PKP1	T
Platelet glutaminase	GLS	T
Platelet monamine oxidase		T
Plectin 1	PLEC1	T
Polycystic kidney and hepatic disease 1	PKHD1	T
Polycystin 1	PKD1	T
Polycystin 2	PKD2	T
Polymorphonuclear elastase		T
Preproglucagon		T
Preproinsulin		T
Presenilin 1	PSEN1	T
Presenilin 2	PSEN2	T
Prostaglandin I2 receptor		T
Protease inhibitor 1		T
Renal glutaminase		T
Retinaldehyde binding protein 1	RLBP1	T
Retinol binding protein 1		T
Retinol binding protein 2		T
Retinol binding protein 4	RBP4	T
Rhesus blood group, CcEe antigens	RHCE	T
Rhesus blood group, D antigen	RHD	T
Rhesus blood group-associated glycoprotein	RHAG	T
Salivary amylase, AMY1		T
Secretin	SCT	T
Secretin receptor, SCTR	SCTR	T
Serum amyloid A	SAA	T
Serum amyloid P	SAP	T
Sex hormone binding globulin, SHBG		T
Solute carrier family 1 (amino acid transporter), member 6	SLC1A6	T
Solute carrier family 1 (glial high affinity glutamate transporter), member 3	SLC1A3	T
Solute carrier family 1 (glutamate transporter), member 1	SLC1A1	T
Solute carrier family 1 (glutamate transporter), member 2	SLC1A2	T
Solute carrier family 1 (neutral amino acid transporter), member 4	SLC1A4	T
Solute carrier family 10 (sodium/bile acid cotransporter family),member 1	SLC10A1	T
Solute carrier family 10 (sodium/bile acid cotransporter family),member 2	SLC10A2	T
Solute carrier family 12, member 1	SLC12A1	T
Solute carrier family 12, member 2	SLC12A2	T

Solute carrier family 12, member 3	SLC12A3	T
Solute carrier family 14, member 2	SLC14A2	T
Solute carrier family 15 (H+/peptide transporter, intestinal), member 1	SLC15A1	T
Solute carrier family 15 (H+/peptide transporter, kidney), member 2	SLC15A2	T
Solute carrier family 16 (monocarboxylate transporter), member 1	SLC16A1	T
Solute carrier family 16 (monocarboxylate transporter), member 7	SLC16A7	T
Solute carrier family 17, member 1	SLC17A1	T
Solute carrier family 17, member 2	SLC17A2	T
Solute carrier family 18, member 3	SLC18A3	T
Solute carrier family 19 (folate transporter), member 1	SLC19A1	T
Solute carrier family 2 (facilitated glucose transporter), member 1	SLC2A1	T
Solute carrier family 2 (facilitated glucose transporter), member 2	SLC2A2	T
Solute carrier family 2 (facilitated glucose transporter), member 3	SLC2A3	T
Solute carrier family 2 (facilitated glucose transporter), member 4	SLC2A4	T
Solute carrier family 2 (facilitated glucose transporter), member 5	SLC2A5	T
Solute carrier family 20, member 1	SLC20A1	T
Solute carrier family 20, member 2	SLC20A2	T
Solute carrier family 20, member 3	SLC20A3	T
Solute carrier family 21, member 2	SLC21A2	T
Solute carrier family 21, member 3	SLC21A3	T
Solute carrier family 22, member 1	SLC22A1	T
Solute carrier family 22, member 2	SLC22A2	T
Solute carrier family 22, member 5	SLC22A5	T
Solute carrier family 25, member 12	SLC25A12	T
Solute carrier family 3 (facilitated glucose transporter), member 1	SLC3A1	T
Solute carrier family 4 (anion exchanger), member 1	SLC4A1	T
Solute carrier family 4 (anion exchanger), member 2	SLC4A2	T
Solute carrier family 4 (anion exchanger), member 3	SLC4A3	T
Solute carrier family 5 (sodium/glucose transporter), member 1	SLC5A1	T
Solute carrier family 5 (sodium/glucose transporter), member 2	SLC5A2	T
Solute carrier family 5 (sodium/glucose transporter), member 5	SLC5A5	T
Solute carrier family 5, member 3	SLC5A3	T
Solute carrier family 6 (GAMMA-	SLC6A1	T

AMINOBUTYRIC ACID transporter), member 1		
Solute carrier family 6 (neurotransmitter transporter, dopamine), member 3	SLC6A3	T
Solute carrier family 6 (neurotransmitter transporter, noradrenaline), member 2	SLC6A2	T
Solute carrier family 6 (neurotransmitter transporter, serotonin), member 4	SLC6A4	T
Solute carrier family 6, member 10	SLC6A10	T
Solute carrier family 6, member 6	SLC6A6	T
Solute carrier family 6, member 8	SLC6A8	T
Solute carrier family 7(amino acid transporter), member 1	SLC7A1	T
Solute carrier family 7(amino acid transporter), member 2	SLC7A2	T
Solute carrier family 7(amino acid transporter), member 7	SLC7A7	T
Solute carrier family 8 (sodium/calcium exchanger), member 1	SLC8A1	T
Sorcin	SRI	T
Steroidogenic acute regulatory protein	STAR	T
Sterol carrier protein 2	SCP2	T
Stratum corneum chymotryptic enzyme		T
Sucrase-isomaltase	SI	T
Surfactant pulmonary-associated protein A1	SFTPA1	T
Surfactant pulmonary-associated protein A2	SFTPA2	T
Surfactant pulmonary-associated protein B	SFTPB	T
Surfactant pulmonary-associated protein C	SFTPC	T
Surfactant pulmonary-associated protein D	SFTPD	T
Survival of motor neuron 1, telomeric	SMN1	T
Tetranectin	TNA	T
Thyroxin-binding globulin	TBG	T
Tocopherol (alpha) transfer protein	TTPA	T
Transcobalamin 1, TCN1		T
Transcobalamin 2, TCN2	TCN2	T
Transthyretin	TTR	T
Trehalase		T
Trypsinogen activation peptide		T
Uncoupling protein 1		T
Uncoupling protein 3	UCP3	T
Uteroglobin	UGB	T
Vitelliform macular dystrophy, atypical gene	VMD1	T
Vitronectin receptor, alpha	VNRA	T
Von Willebrand factor	VWF	T
Achromatopsia 2	ACHM2	S
Actin, alpha, skeletal	ACTA1	S
Actin, alpha, smooth, aortic	ACTA2	S
Actin, alpha, cardiac	ACTC	S
Actin, beta	ACTB	S
Actin, gamma 2	ACTG2	S
Adducin, alpha	ADD1	S

Adducin, beta	ADD2	S
Amelogenin	AMELX	S
Ankyrin 1	ANK1	S
Ankyrin 2	ANK2	S
Ankyrin 3	ANK3	S
Apaf-1		S
Arrestin	SAG	S
Blue cone pigment	BCP	S
Chloride channel 1, skeletal muscle	CLCN1	S
Chloride channel 5	CLCN5	S
Chloride channel KB	CLCNKB	S
Choroideremia gene	CHM	S
Cofilin		S
Collagen I alpha 1	COL1A1	S
Collagen I alpha 2	COL1A2	S
Collagen II alpha 1	COL2A1	S
Collagen III alpha 1	COL3A1	S
Collagen IV alpha 1	COL4A1	S
Collagen IV alpha 2	COL4A2	S
Collagen IV alpha 3	COL4A3	S
Collagen IV alpha 4	COL4A4	S
Collagen IV alpha 5	COL4A5	S
Collagen IV alpha 6	COL4A6	S
Collagen IX alpha 2	COL9A2, EDM2	S
Collagen IX alpha 3	COL9A3	S
Collagen receptor	COLR	S
Collagen V alpha 1	COL5A1	S
Collagen V alpha 2	COL5A2	S
Collagen VI alpha 1	COL6A1	S
Collagen VI alpha 2	COL6A2	S
Collagen VI alpha 3	COL6A3	S
Collagen VII alpha 1	COL7A1	S
Collagen X alpha 1	COL10A1	S
Collagen X alpha 1	COL11A1	S
Collagen XI alpha 2	COL11A2	S
Collagen XVII alpha 1	COL17A1	S
Cryptochrome 1	CRY1	S
Cryptochrome 2	CRY2	S
Crystallin, alpha A	CRYAA	S
Crystallin, alpha B	CRYAB	S
Crystallin, beta B2	CRYBB2	S
Crystallin, gamma A	CRYGA	S
Desmin	DES	S
DNA damage binding protein, DDB1	DDB1	S
DNA damage binding protein, DDB2	DDB2	S
DNA-damage-inducible transcript 3	DDIT3	S
Doublecortin, DCX	DCX	S
Dyskerin	DKC1	S
Dystonia 1	DYT1	S
Dystonia 3	DYT3	S

Dystonia 6	DYT6	S
Dystonia 7	DYT7	S
Dystonia 9	CSE	S
Dystrophin	DMD	S
Dystrophin-associated glycoprotein 35kD, SCGD	SGCD	S
Dystrophin-associated glycoprotein 35kD, SGSG	SGCG	S
Dystrophin-associated glycoprotein 43kD	SGCB	S
Dystrophin-associated glycoprotein 50kD	SGCA	S
Ectodermal Dysplasia 1 gene	ED1	S
Elastin	ELN	S
Endocardial fibroelastosis 2 gene	EFE2	S
Endoglin	ENG	S
Erythrocyte membrane protein band 4.1	EPB41	S
Erythrocyte membrane protein band 4.2	EPB42	S
Erythrocyte membrane protein band 7.2	EPB72	S
Exostosin 1	EXT1	S
Exostosin 2	EXT2	S
Exostosin 3	EXT3	S
Eye colour gene 3 (brown)	EYCL3	S
Fibrinogen alpha	FGA	S
Fibrinogen beta	FGB	S
Fibrinogen gamma	FGG	S
Glycophorin A	GYPA	S
Glycophorin B	GYPB	S
Glycophorin C	GYPC	S
Green cone pigment	GCP	S
Keratin 1	KRT1	S
Keratin 10	KRT10	S
Keratin 11	KRT11	S
Keratin 12	KRT12	S
Keratin 13	KRT13	S
Keratin 14	KRT14	S
Keratin 15	KRT15	S
Keratin 16	KRT16	S
Keratin 17	KRT17,PCHC1	S
Keratin 18	KRT18	S
Keratin 2	KRT2	S
Keratin 3	KRT3	S
Keratin 4	KRT4	S
Keratin 5	KRT5	S
Keratin 6	KRT6	S
Keratin 7	KRT7	S
Keratin 8	KRT8	S
Keratin 9	KRT9	S
Keratin, hair acidic 1	KRTHA1	S
Keratin, hair basic 2	KRTHB1	S
Keratin, hair basic 6	KRTHB6	S
Loricrin	LOR	S
Microtuble associated protein	MAP	S
Moesin, MSN		S

Myomesin 1	MYOM1	S
Myomesin 2	MYOM2	S
Myelin basic protein		S
Myelin protein peripheral 22	PMP22	S
Myelin protein zero	MPZ	S
Myosin 15	MYO15	S
Myosin 5A	MYO5A	S
Myosin 6	MYO6	S
Myosin 7A	MYO7A	S
Myosin, cardiac	MYH7	S
Myosin, light chain 2	MYL2	S
Myosin, light chain 3	MYL3	S
Myosin-binding protein C, cardiac	MYBPC3	S
Myotubularin	MTM1	S
Nebulin	NEB	S
Neurofilament protein, heavy	NFH	S
Neurofilament protein, NF125	NF150	S
Neurofilament protein, NF200	NF200	S
Neurofilament protein, NF68	NF68	S
Ocular albinism 1	OA1	S
Oculocutaneous albinism II	OCA2	S
Osteocalcin		S
Peripherin, PRPH		S
Peroxisomal membrane protein 1	PXMP1	S
Persyn		S
Proline-rich protein BstNI subfamily 1	PRB1	S
Proline-rich protein BstNI subfamily 3	PRB3	S
Proline-rich protein BstNI subfamily 4	PRB4	S
Radixin	RDX	S
Red cone pigment	RCP	S
Retinal pigment epithelium specific protein (65kD)	RPE65	S
Retinitis pigmentosa gene 1	RP1	S
Retinitis pigmentosa gene 2	RP2	S
Retinitis pigmentosa gene 3	RP3	S
Retinitis pigmentosa gene 6	RP6	S
Retinitis pigmentosa gene 7	RP7, RDS	S
Rhodopsin	RHO	S
Rod outer segment membrane protein 1	ROM1	S
Semaphorin A4	SEMA4	S
Semaphorin A5	SEMA5	S
Semaphorin D		S
Semaphorin E	SEMAE	S
Semaphorin F	SEMA3/F	S
Semaphorin W	SEMAW	S
Small nuclear ribonucleoprotein polypeptide N	SNRPN	S
Spectrin alpha	SPTA1	S
Spectrin beta	SPTB	S
Talin, TLN		S
Tau protein	MAPT	S
Tenascin (cytotactin)		S

Tenascin XA	TNXA	S
Titin	TTN	S
Tropomyosin 1 alpha	TPM1	S
Tropomyosin 3 (non-muscle)	TPM3	S
Troponin C		S
Troponin I	TNNI3	S
Troponin T2, cardiac	TNNT2	S
Tubulin		S
Undulin 1	COL14A1	S
Usher syndrome 2A	USH2A	S
Villin		S
Vinculin		S
Wolfram syndrome 1 gene	WFS1	S
Zinc finger protein 198	ZIC198	S
Zinc finger protein 2	ZIC2	S
Zinc finger protein 3	ZIC3	S
Zinc finger protein HRX	ALL1	I
Alpha 2 macroglobulin	A2M	I
Annexin 1	ANX 1	I
Apoptosis antigen 1	APT1	I
Apoptosis antigen ligand 1	APT1LG1	I
Apoptosis-inducing factor	AIF	I
ATP-binding cassette transporter 7	ABC7	I
Attractin		I
Autoimmune regulator, AIRE	AIRE	I
B-cell CLL/lymphoma 1	BCL1	I
B-cell CLL/lymphoma 10	BCL10	I
B-cell CLL/lymphoma 3	BCL3	I
B-cell CLL/lymphoma 4	BCL4	I
B-cell CLL/lymphoma 5	BCL5	I
B-cell CLL/lymphoma 6	BCL6	I
B-cell CLL/lymphoma 7	BCL7	I
B-cell CLL/lymphoma 8	BCL8	I
B-cell CLL/lymphoma 9	BCL9	I
beta 2 microglobulin	B2M	I
Bradykinin receptor B1		I
Bradykinin receptor B2		I
Calcineurin A1	CALNA1	I
Calcineurin A2	CALNA2	I
Calcineurin A3	CALNA3	I
Calcineurin B		I
Catalase	CAT	I
CD1	CD1	I
CD10	CD10	I
CD100	CD100	I
CD101	CD101	I
CD103	CD103	I
CD106	CD106	I
CD107	CD107	I
CD108	CD108	I

CD109	CD109 I
CD110	CD110 I
CD111	CD111 I
CD112	CD112 I
CD113	CD113 I
CD114	CD114 I
CD115	CD115 I
CD116	CD116 I
CD117	CD117 I
CD118	CD118 I
CD119	CD119 I
CD12	CD12 I
CD120	CD120 I
CD121	CD121 I
CD122	CD122 I
CD123	CD123 I
CD124	CD124 I
CD125	CD125 I
CD126	CD126 I
CD127	CD127 I
CD128	CD128 I
CD129	CD129 I
CD13	CD13 I
CD130	CD130 I
CD131	CD131 I
CD132	CD132 I
CD133	CD133 I
CD134	CD134 I
CD135	CD135 I
CD136	CD136 I
CD137	CD137 I
CD138	CD138 I
CD139	CD139 I
CD14	CD14 I
CD140	CD140 I
CD141	CD141 I
CD142	CD142 I
CD143	CD143 I
CD144	CD144 I
CD145	CD145 I
CD147	CD147 I
CD148	CD148 I
CD149	CD149 I
CD15	CD15 I
CD150	CD150 I
CD151	CD151 I
CD152	CD152 I
CD153	CD153 I
CD154	CD154 I
CD155	CD155 I

CD156	CD156 I
CD157	CD157 I
CD158	CD158 I
CD159	CD159 I
CD160	CD160 I
CD161	CD161 I
CD162	CD162 I
CD163	CD163 I
CD164	CD164 I
CD165	CD165 I
CD166	CD166 I
CD17	CD17 I
CD19	CD19 I
CD2	CD2 I
CD20	CD20 I
CD22	CD22 I
CD23	CD23 I
CD24	CD24 I
CD25	CD25 I
CD26	CD26 I
CD27	CD27 I
CD28	CD28 I
CD3	CD3 I
CD30	CD30 I
CD31	CD31 I
CD33	CD33 I
CD34	CD34 I
CD36	CD36 I
CD37	CD37 I
CD38	CD38 I
CD39	CD39 I
CD4	CD4 I
CD40	CD40 I
CD41	CD41 I
CD42	CD42 I
CD43	CD43 I
CD44	CD44 I
CD45	CD45 I
CD46	CD46 I
CD47	CD47 I
CD48	CD48 I
CD5	CD5 I
CD50	CD50 I
CD52	CD52 I
CD53	CD53 I
CD55	CD55 I
CD57	CD57 I
CD58	CD58 I
CD59	CD59 I
CD6	CD6 I

CD60	CD60	I
CD63	CD63	I
CD65	CD65	I
CD66	CD66	I
CD67	CD67	I
CD68	CD68	I
CD69	CD69	I
CD7	CD7	I
CD70	CD70	I
CD71	CD71	I
CD72	CD72	I
CD73	CD73	I
CD74	CD74	I
CD75	CD75	I
CD76	CD76	I
CD77	CD77	I
CD78	CD78	I
CD79	CD79	I
CD8	CD8	I
CD80	CD80	I
CD81	CD81	I
CD83	CD83	I
CD84	CD84	I
CD85	CD85	I
CD86	CD86	I
CD88	CD88	I
CD89	CD89	I
CD9	CD9	I
CD90	CD90	I
CD91	CD91	I
CD92	CD92	I
CD93	CD93	I
CD94	CD94	I
CD96	CD96	I
CD97	CD97	I
CD98	CD98	I
CD99	CD99	I
Chemokine MCAF	MCAF	I
Chemokine receptor CCR2	CCR2	I
Chemokine receptor CCR3	CCR3	I
Chemokine receptor CCR5	CCR5	I
Chemokine receptor CXCR1	CXCR1	I
Chemokine receptor CXCR2	CXCR2	I
Chemokine receptor CXCR4	CXCR4	I
Cholesterylester hydrolase		I
Chondritin Sulphate A - placental receptor		I
Cochlin	COCH	I
Complement component C1 inhibitor	C1NH	I
Complement component C1qa	C1QA	I
Complement component C1qb	C1QB	I

Complement component C1qg	C1QG	I
Complement component C1r	C1R	I
Complement component C1s	C1S	I
Complement component C2	C2	I
Complement component C3	C3	I
Complement component C4A	C4A	I
Complement component C4B	C4B	I
Complement component C5	C5	I
Complement component C6	C6	I
Complement component C7	C7	I
Complement component C8	C8B	I
Complement component C9	C9	I
Complement component receptor 1	CR1	I
Complement component receptor 2	CR2	I
Complement component receptor 3	CR3	I
Corticosteroid nuclear receptor		I
Cortisol receptor		I
C-reactive protein CRP		I
Cyclophilin		I
Cytokine-suppressive antiinflammatory drug-binding protein 1	CSBP1	I
Cytokine-suppressive antiinflammatory drug-binding protein 2	CSBP2	I
DAX1 nuclear receptor	DAX1	I
Endo-P-D-glucuronidase		I
Erythropoietin	EPO	I
Erythropoietin receptor	EPOR	I
Factor 1 (No. one)	F1	I
Factor B, properdin		I
Factor D		I
Factor H	HF1	I
Factor I (letter I)	IF	I
Factor III	F3	I
Factor IX	F9	I
Factor V	F5	I
Factor VII	F7	I
Factor VIII	F8	I
Factor X	F10	I
Factor XI	F11	I
Factor XII	F12	I
Factor XIII A & B	F13A & F13B	I
Fc receptor		I
Follicular lymphoma variant translocation 1	FVT1	I
Gastrointestinal tumor-associated antigen 1	GA733	I
Growth-regulated protein precursor, GRO	GRO	I
Haptoglobin, alpha 1	HPA1	I
Haptoglobin, alpha 2	HPA2	I
Haptoglobin, beta	HPB	I
Heat shock protein, HSP60		I
Heat shock protein, HSP70		I

Heat shock protein, HSP90		I
Heat shock protein, HSPA1		I
Heat shock protein, HSPA2		I
Hemopexin	HPX	I
Heparin Cofactor II	HCF2	I
Hepatitis B virus integration site 1	HVBS1	I
Hepatitis B virus integration site 2	HVBS6	I
Histatin 1		I
Histatin 2		I
Histatin 3	HTN3	I
HLA-B associated transcript 1	BAT1	I
IC7 A and B		I
Immunoglobulin alpha (IgA)	IGHA	I
Immunoglobulin gamma (IgG) 2	IGHG2	I
Immunoglobulin delta (IgD)	IGHD	I
Immunoglobulin epsilon (IgE)	IGHE	I
Immunoglobulin E (IgE) reponsiveness gene	IGER	I
Immunoglobulin E (IgE) serum concentration regulator gene	IGES	I
Immunoglobulin heavy mu chain	IGHM	I
Immunoglobulin J polypeptide	IGJ	I
Immunoglobulin kappa constant region	IGKC	I
Immunoglobulin kappa variable region	IGKV	I
Intercellular adhesion molecule 1	ICAM1	I
Intercellular adhesion molecule 2	ICAM2	I
Intercellular adhesion molecule 3	ICAM3	I
Interferon alpha	IFNA1	I
Interferon beta	IFNB	I
Interferon gamma	IFNG	I
Interferon gamma receptor 1	IFNGR1	I
Interferon gamma receptor 2	IFNGR2	I
Interferon regulatory factor 1	IRF1	I
Interferon regulatory factor 4	IRF4	I
Interleukin(IL) 1 receptor	IL1R	I
Interleukin(IL) 1, alpha	IL1A	I
Interleukin(IL) 1, beta	IL1B	I
Interleukin(IL) 10	IL10	I
Interleukin(IL) 10 receptor	IL10R	I
Interleukin(IL) 11	IL11	I
Interleukin(IL) 11 receptor	IL11R	I
Interleukin(IL) 12	IL12	I
Interleukin(IL) 12 receptor, beta 1	IL12RB1	I
Interleukin(IL) 13	IL13	I
Interleukin(IL) 13 receptor	IL13R	I
Interleukin(IL) 2	IL2	I
Interleukin(IL) 2 receptor, alpha	IL2RA	I
Interleukin(IL) 2 receptor, gamma	IL2RG	I
Interleukin(IL) 3	IL3	I
Interleukin(IL) 3 receptor	IL3R	I
Interleukin(IL) 4	IL4	I

Interleukin(IL) 4 receptor	IL4R	I
Interleukin(IL) 5	IL5	I
Interleukin(IL) 5 receptor	IL5R	I
Interleukin(IL) 6	IL6	I
Interleukin(IL) 6 receptor	IL6R	I
Interleukin(IL) 7	IL7	I
Interleukin(IL) 7 receptor	IL7R	I
Interleukin(IL) 8	IL8	I
Interleukin(IL) 8 receptor	IL8R	I
Interleukin(IL) 9	IL9	I
Interleukin(IL) 9 receptor	IL9R	I
Interleukin(IL) receptor antagonist 1	IL1RN, IL1RA	I
Kallikrein 3	KAK3	I
Kininogen, High molecular weight	KNG	I
Lectin, mannose-binding 1	LMAN1	I
Lectin, mannose-binding 2	MBL2	I
Leukin		I
Leukocyte-specific transcript 1	LST-1	I
Leukotriene A4 hydrolase		I
Leukotriene B4 receptor		I
Leukotriene C4 receptor		I
Leukotriene D4/E4 receptor		I
LIM-Kinase I (LINK-I)		I
Lipocortin 1	ANX4	I
Lipoprotein lipase	LPL	I
Lipoprotein-associated coagulation factor	LACI	I
Lipoxygenase 12 (platelets)	LOG12	I
Lipoxygenase 5 (leukocytes)		I
Lymphoblastic leukemia derived sequence 1	LYL1	I
Lymphocyte-specific protein tyrosine kinase	LCK	I
lymphotoxin		I
Lysozyme	LYZ	I
Macrophage activating factor	MAF	I
Macrophage inflammatory protein-1	MIP1	I
Macrophage inflammatory protein-1 receptor		I
Macrophage inflammatory protein-2	MIP2	I
Macrophage inflammatory protein-2 receptor		I
Malignant proliferation, eosinophil gene	MPE	I
Mannose binding protein	MBP	I
MHC Class I: A		I
MHC Class I: B		I
MHC Class I: C		I
MHC Class I: LMP-2, LMP-7		I
MHC Class I: Tap1	ABCR, TAP1	I
MHC Class II: DP	HLA-DPB1	I
MHC Class II: DQ		I
MHC Class II: DR		I
MHC Class II: Tap2	TAP2, PSF2	I
MHC Class II:Complementation group A	MHC2TA	I
MHC Class II:Complementation group B	rfxank	I

MHC Class II:Complementation group C	RFX5	I
MHC Class II:Complementation group D	RFXAP	I
Monocyte chemoattractant protein 1	MCP1	I
Myeloid leukemia factor-1	MLF1	I
Myeloperoxidase	MPO	I
N-acyl hydrolase		I
NADPH oxidase		I
Natural resistance-associated macrophage protein 1	NRAMP1	I
NB6		I
Neuronal apoptosis inhibitory protein	NAIP	I
Neuronal molecule-1		I
Neuronal molecule-1 receptor		I
Neutrophil cystolic factor 1	NCF1	I
Neutrophil cystolic factor 2	NCF2	I
Nuclear factor I-kappa-B-like gene	IKBL	I
Nuclear factor kappa beta	NFKB	I
Peanut-like 1	PNUTL1	I
Phagocytin		I
Phospholipase A2, group 10	PLA2G10	I
Phospholipase A2, group 1B	PLA2G1B	I
Phospholipase A2, group 2A	PLA2G2A	I
Phospholipase A2, group 2B	PLA2G2B	I
Phospholipase A2, group 4A	PLA2G4A	I
Phospholipase A2, group 4C	PLA2G4C	I
Phospholipase A2, group 5	PLA2G5	I
Phospholipase A2, group 6	PLA2G6	I
Phospholipase C alpha		I
Phospholipase C beta		I
Phospholipase C delta	PLCD1	I
Phospholipase C epsilon		I
Phospholipase C gamma	PLCG1	I
Platelet glycoprotein 1b, alpha	GP1BA	I
Platelet glycoprotein 1b, beta	GP1BB	I
Platelet glycoprotein 1b, gamma	GP1BG	I
Platelet glycoprotein IX	GP9	I
Platelet glycoprotein V	GP5	I
Platelet-activating factor acetylhydrolase 1B	PAFAH1B1 or LIS1	I
Platelet-activating factor acetylhydrolase 2	PAFAH2	I
Platelet-activating factor receptor	PAFR	I
Poliovirus receptor	PVR, PVS	I
Prekallikrein		I
Properdin P factor, complement	PFC, PFD	I
Prostacyclin synthase		I
Prostaglandin 15-OH dehydrogenase	HGPD; PGDH	I
Prostaglandin D - DP receptor		I
Prostaglandin E1 receptor		I
Prostaglandin E2 receptor		I
Prostaglandin E3 receptor		I
Prostaglandin F - FP receptor		I
Prostaglandin F2 alpha receptor		I

Prostaglandin IP receptor		I
Protein C	PROC	I
Protein C inhibitor	PCI	I
Protein S	PROS1	I
Proteinase 3		I
Prothrombin precursor	F2	I
SAP (SLAM-associated protein)	SH2D1A	I
Severe combined immunodeficiency, type A (Athabascan)	SCIDA	I
Signaling lymphocyte activation molecule	SLAM	I
Sjoegren (Sjogren) syndrome antigen A1	SSA1	I
SYK-related tyrosine kinase	SRK	I
T-cell acute lymphocytic leukemia 1	TAL1	I
T-cell acute lymphocytic leukemia 2	TAL2	I
T-cell receptor, alpha	TCRA	I
T-cell receptor, delta	TCRD	I
Terminal deoxynucleotidyltransferase	TDT	I
Thrombin receptor	F2R	I
Thrombomodulin	THBD	I
Thromboxane A synthase 1	TBXAS1	I
Thromboxane A2	TXA2	I
Thromboxane A2 receptor	TBXA2R	I
Thy-1 T-cell antigen	THY1	I
Thymic humoral factor		I
Thymosin		I
Tip-associated protein	TAP	I
Toll-like receptor 4	TLR4	I
Tumour necrosis factor (TNF) receptor associated factor 1	TRAF1	I
Tumour necrosis factor (TNF) receptor associated factor 2	TRAF2	I
Tumour necrosis factor (TNF) receptor associated factor 3	TRAF3	I
Tumour necrosis factor (TNF) receptor associated factor 4	TRAF4	I
Tumour necrosis factor (TNF) receptor associated factor 5	TRAF5	I
Tumour necrosis factor (TNF) receptor associated factor 6	TRAF6	I
Tumour necrosis factor alpha	TNFA	I
Tumour necrosis factor alpha receptor	TNFAR	I
Tumour necrosis factor beta	TNFB	I
Tumour necrosis factor beta receptor	TNFBR	I
Tumour suppresssor gene DRA	DRA	I
Uridine monophosphate kinase	UMPK	I
Uridine monophosphate synthetase	UMPS	I
Vimentin	VIM	I
Wiskott-Aldrich syndrome protein	WASP, THC	I
17-ketosteroid reductase		N
Acetylcholine receptor, nicotinic, alpha A1	CHRNA1	N

Acetylcholine receptor, nicotinic, alpha A2	CHRNA2	N
Acetylcholine receptor, nicotinic, alpha A3	CHRNA3	N
Acetylcholine receptor, nicotinic, alpha A4	CHRNA4	N
Acetylcholine receptor, nicotinic, alpha A5	CHRNA5	N
Acetylcholine receptor, nicotinic, alpha A6	CHRNA6	N
Acetylcholine receptor, nicotinic, alpha A7	CHRNA7	N
Acetylcholine receptor, nicotinic, beta 1	CHRNB1	N
Acetylcholine receptor, nicotinic, beta 2	CHRNB2	N
Acetylcholine receptor, nicotinic, beta 3	CHRNB3	N
Acetylcholine receptor, nicotinic, beta 4	CHRNB4	N
Acetylcholine receptor, nicotinic, epsilon	CHRNE	N
Acetylcholine receptor, nicotinic, gamma	CHRNG	N
Adenosine receptor A1	ADORA1	N
Adenosine receptor A2A	ADORA2A	N
Adenosine receptor A2B	ADORA2B	N
Adenosine receptor A3	ADORA3	N
Adenyl cyclase		N
Adrenergic receptor, alpha1	ADRA1	N
Adrenergic receptor, alpha2	ADRA2	N
Adrenergic receptor, beta1	ADRB1	N
Adrenergic receptor, beta2	ADRB2	N
Adrenergic receptor, beta3	ADRB3	N
alpha thalassemia gene	ATRX	N
alpha-synuclein	SNCA	N
Amyloid beta (A4) precursor protein-binding, APBB1	APBB1	N
Amyloid beta A4 precursor protein	APP	N
Amyloid beta A4 precursor-like protein	APLP	N
Arginine vasopressin	AVP	N
Arginine vasopressin receptor 1A	AVPR1A	N
Arginine vasopressin receptor 1B	AVPR1B	N
Arginine vasopressin receptor 2	AVPR2	N
Aspartate receptor		N
Benzodiazepine receptor		N
beta-endorphin receptor		N
beta-synuclein	SNCB	N
Calcitonin receptor /Calcitonin gene-related peptide receptor	CALCR	N
Calcitonin/Calcitonin gene-related peptide alpha	CALCA	N
Calcium channel, voltage-dependent, alpha 1F subunit	CACNA1F	N
Calcium channel, voltage-dependent, Alpha-1B (CACNL1A5)	CACNA1B	N
Calcium channel, voltage-dependent, Alpha-1C	CACNA1C	N
Calcium channel, voltage-dependent, Alpha-1D	CACNA1D	N
Calcium channel, voltage-dependent, Alpha-1E (CACNL1A6)	CACNA1E	N
Calcium channel, voltage-dependent, Alpha-2/delta	CACNA2	N
Calcium channel, voltage-dependent, Beta 1	CACNB1	N
Calcium channel, voltage-dependent, Beta 3	CACNB3	N

Calcium channel, voltage-dependent, L type, alpha 1S subunit	CACNA1S	N
Calcium channel, voltage-dependent, Neuronal, Gamma	CACNG2	N
Calcium channel, voltage-dependent, P/Q type, alpha 1A subunit	CACNA1A	N
Calcium channel, voltage-dependent, T-type		N
Calretinin	CALB2	N
Cannabinoid receptor	CNR1	N
Carnosinase		N
Cartilage oligomeric matrix protein	COMP, EDM1, PSACH	N
Cartilage-hair hypoplasia gene	CHH	N
Cellubrevin	CEB	N
Ceroid lipofuscinosis neuronal 2	CLN2	N
Ceroid lipofuscinosis neuronal 3	CLN3	N
Ceroid lipofuscinosis neuronal 4	CLN4	N
Ceroid lipofuscinosis neuronal 5	CLN5	N
Ceroid lipofuscinosis neuronal 6	CLN6	N
Cholecystokinin	CCK	N
Cholecystokinin B receptor	CCKBR	N
Corticosteroid binding globulin	CBG	N
Cyclic nucleotide gated channel alpha 1, CNGA1	CNGA1	N
Cyclic nucleotide gated channel alpha 3, CNGA3	CNGA3	N
Cystic fibrosis transmembrane conductance regulator, CFTR	CFTR	N
Deafness autosomal dominant 5	DFNA5	N
Deafness dystonia peptide	DDP	N
Diaphanous 1	DIAPH1	N
Diaphanous 2	DIAPH2	N
Dihydrolipoamide branched chain transacylase	DBT	N
Dihydrolipoamide dehydrogenase	DLD	N
Dihydrolipoamide succinyltransferase		N
Dopamine receptors D1	DRD1	N
Dopamine receptors D2	DRD2	N
Dopamine receptors D3	DRD3	N
Dopamine receptors D4	DRD4	N
Dopamine receptors D5	DRD5	N
Dynorphin receptor		N
Endobrevin	VAMP8	N
Endothelin 1	EDN1	N
Endothelin 2	EDN2	N
Endothelin 3	EDN3	N
Endothelin converting enzyme	ECE1	N
Endothelin receptor type A	EDNRA	N
Endothelin receptor type B	EDNRB	N
Fragile site, folic acid type, rare, fra(X) A	FRAXA	N
Fragile site, folic acid type, rare, fra(X) E	FRAXE	N
Fragile site, folic acid type, rare, fra(X) F	FRAXF	N
GABA receptor, alpha 1	GABRA1	N

GABA receptor, alpha 2	GABRA2	N
GABA receptor, alpha 3	GABRA3	N
GABA receptor, alpha 4	GABRA4	N
GABA receptor, alpha 5	GABRA5	N
GABA receptor, alpha 6	GABRA6	N
GABA receptor, beta 1	GABRB1	N
GABA receptor, beta 2	GABRB2	N
GABA receptor, beta 3	GABRB3	N
GABA receptor, gamma 1	GABRG1	N
GABA receptor, gamma 2	GABRG2	N
GABA receptor, gamma 3	GABRG3	N
Galanin	GAL	N
Galanin receptor	GALNR1	N
Gephyrin		N
Glial-cell derived neurotrophic factor (GDNF) receptor		N
Glial-cell derived neurotrophic factor, GDNF	GDNF	N
Glutamate receptor 1	GLUR1	N
Glutamate receptor 2	GLUR2	N
Glutamate receptor 3	GLUR3	N
Glutamate receptor 4	GLUR4	N
Glutamate receptor 5	GLUR5	N
Glutamate receptor 6	GLUR6	N
Glutamate receptor 7	GLUR7	N
Glutamate receptor, ionotropic, NMDA 1	NMDAR1	N
Glutamate receptor, ionotropic, NMDA 2A	NMDAR2A	N
Glutamate receptor, ionotropic, NMDA 2B	NMDAR2B	N
Glutamate receptor, ionotropic, NMDA 2C	NMDAR2C	N
Glutamate receptor, ionotropic, NMDA 2D	NMDAR2D	N
Glycine receptor, alpha	GLRA2	N
Glycine receptor, beta		N
Glycine transporter	GLYT	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 1, GNAI1	GNAI1	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 2, GNAI2	GNAI2	N
Guanine nucleotide-binding protein, alpha inhibiting activity polypeptide 3, GNAI3	GNAI3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS1	GNAS1	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS2	GNAS2	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS3	GNAS3	N
Guanine nucleotide-binding protein, alpha stimulating activity polypeptide, GNAS4	GNAS4	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT1	GNAT1	N
Guanine nucleotide-binding protein, alpha transducing activity polypeptide, GNAT2	GNAT2	N

Guanine nucleotide-binding protein, alpha activating activity polypeptide, GNAO	GNAO1	N
Guanine nucleotide-binding protein, beta polypeptide 3	GNB3	N
Guanine nucleotide-binding protein, gamma polypeptide 5	GNG5	N
Guanine nucleotide-binding protein, q polypeptide	GNAQ	N
Gustducin, alpha (taste-specific G protein)	GDCA	N
H(+), K(+) - ATPase	ATP4B	N
Hippocampal cholinergic neurostimulating peptide,	HCNP	N
Histamine receptors, H1		N
Histamine receptors, H2		N
Histamine receptors, H3		N
Inositol monophosphatase	IMPA1	N
Inositol polyphosphate 1-phosphatase	INPP1	N
Islet amyloid polypeptide	IAPP	N
L1 cell adhesion molecule	L1CAM	N
Luteinizing hormone-releasing hormone		N
Luteinizing hormone-releasing hormone receptor		N
Melatonin receptor 1A	MTNR1A	N
Melatonin receptor 1B	MTNR1B	N
Muscarinic receptor, M1	CHRM1	N
Muscarinic receptor, M2	CHRM2	N
Muscarinic receptor, M3	CHRM3	N
Muscarinic receptor, M4	CHRM4	N
Muscarinic receptor, M5	CHRM5	N
Neurexin		N
Neurite growth-promoting factor 2	MDK	N
Neurite inhibitory protein		N
Neurokinin A	NKNA	N
Neurokinin B	NKNB	N
Neuropeptide Y	NPY	N
Neuropeptide Y receptor Y1	NPY1R	N
Neuropeptide Y receptor Y2	NPY2R	N
Neurotensin	NTS	N
Neurotensin receptor	NTSR1	N
Opioid receptor, delta	OPRD1	N
Opioid receptor, kappa	OPRK1	N
Opioid receptor, mu	OPRM1	N
Otoferlin	OTOF	N
Oxytocin	OXT	N
Oxytocin receptor	OXTR	N
Parkin	PARK2	N
Pituitary adenylate cyclase activating peptide	PACAP	N
Pituitary adenylate cyclase activating peptide receptor	PACAP1R	N
Postsynaptic density-95 protein	PSD95	N
Potassium inwardly-rectifying channel J1	KCNJ1	N
Potassium inwardly-rectifying channel J11	KCNJ11	N
Potassium voltage-gated channel A1	KCNA1	N

Potassium voltage-gated channel E1	KCNE1	N
Potassium voltage-gated channel Q1	KCNQ1	N
Potassium voltage-gated channel Q2	KCNQ2	N
Potassium voltage-gated channel Q3	KCNQ3	N
Potassium voltage-gated channel Q4	KCNQ4	N
Potassium channel, subfamily K, member 1	KCNK1	N
Potassium channel, subfamily K, member 2	KCNK2	N
Potassium channel, subfamily K, member 3	KCNK3	N
Potassium channel, calcium-activated,	KCNN4	N
Preproenkephalin	PENK	N
Prion protein	PRNP	N
Prodynorphin		N
Proopiomelanocortin	POMC	N
Prosaposin	PSAP	N
Proteolipid protein	PLP	N
Purinergic receptor P1A1		N
Purinergic receptor P1A2		N
Purinergic receptor P1A3		N
Purinergic receptor P2X, 1	P2RX1	N
Purinergic receptor P2X, 2	P2RX2	N
Purinergic receptor P2X, 3	P2RX3	N
Purinergic receptor P2X, 4	P2RX4	N
Purinergic receptor P2X, 5	P2RX5	N
Purinergic receptor P2X, 6	P2RX6	N
Purinergic receptor P2X, 7	P2RX7	N
Purinergic receptor P2Y, 1	P2RY1	N
Purinergic receptor P2Y, 2	P2RY2	N
Purinergic receptor P2Y, 11	P2RY11	N
Rabphilin		N
RAS-associated protein, RAB3A	RAB3A	N
Rim		N
S100 calcium-binding protein A1	S100A1	N
S100 calcium-binding protein A2	S100A2	N
S100 calcium-binding protein A3	S100A3	N
S100 calcium-binding protein A4	S100A4	N
S100 calcium-binding protein A5	S100A5	N
S100 calcium-binding protein A6	S100A6	N
S100 calcium-binding protein A7	S100A7	N
S100 calcium-binding protein A8	S100A8	N
S100 calcium-binding protein A9	S100A9	N
S100 calcium-binding protein B	S100B	N
S100 calcium-binding protein P	S100P	N
Secretase, alpha		N
Secretase, beta		N
Secretase, gamma		N
Selectin E	SELE	N
Selectin L	SELL	N
Selectin P	SELP	N
Serotonin receptor, 5HT1A	HTR1A	N
Serotonin receptor, 5HT1B	HTR1B	N

Serotonin receptor, 5HT1C	HTR1C	N
Serotonin receptor, 5HT1D	HTR1D	N
Serotonin receptor, 5HT1E	HTR1E	N
Serotonin receptor, 5HT1F	HTR1F	N
Serotonin receptor, 5HT2A	HTR2A	N
Serotonin receptor, 5HT2B	HTR2B	N
Serotonin receptor, 5HT2C	HTR2C	N
Serotonin receptor, 5HT3	HTR3	N
Serotonin receptor, 5HT4	HTR4	N
Serotonin receptor, 5HT5	HTR5	N
Serotonin receptor, 5HT6	HTR6	N
Serotonin receptor, 5HT7	HTR7	N
Sodium channel, non-voltage gated 1, alpha	SCNN1A	N
Sodium channel, non-voltage gated 1, beta	SCNN1B	N
Sodium channel, non-voltage gated 1, gamma	SCNN1G	N
Sodium channel, voltage gated, type IV, alpha polypeptide	SCN4A	N
Sodium channel, voltage gated, type V, alpha polypeptide	SCN5A	N
Sodium channel, voltage-gated, type 1, beta polypeptide	SCN1B	N
Somatostatin	SST	N
Somatostatin receptor, SSTR1	SSTR1	N
Somatostatin receptor, SSTR2	SSTR2	G
Somatostatin receptor, SSTR3	SSTR3	N
Somatostatin receptor, SSTR4	SSTR4	N
Somatostatin receptor, SSTR5	SSTR5	N
Spinocerebellar ataxia 8 gene	SCA8	N
Substance P		N
Synapsin 1a & 1b	SYN1	N
Synapsin 2a & 2b	SYN2	N
Synaptic vesicle amine transporter	SVAT	N
Synaptic vesicle protein 2	SV2	N
Synaptobrevin 1	SYB1	N
Synaptobrevin 2	SYB2	N
Synaptogyrin		N
Synaptophysin	SYP	N
Synaptosomal-associated protein, 25KD	SNAP25	N
Synaptotagmin 1	SYT1	N
Synaptotagmin 2	SYT2	N
Syntaxin 1	STX1	N
Tachykinin receptor, NK1R	TACR1	N
Tachykinin receptor, NK2R	TACR2	N
Tachykinin receptor, NK3R	TACR3	N
Thyrotropin releasing hormone	TRH	N
Thyrotropin releasing hormone receptor	TRHR	N
Transcription factor, TUPLE1	TUPLE1	N
Tremor, essential 1	ETM1	N
Tremor, essential 2	ETM2	N
Tryptophan 2,3-dioxygenase	TDO2	N

Vacuolar proton pump, subunit 1	VPP1	N
Vacuolar proton pump, subunit 3	VPP3	N
Vasoactive intestinal polypeptide	VIP	N
Vasoactive intestinal polypeptide receptor	VIPR	N
Vesicular monoamine transporter 1	VMAT1	N
Vesicular monoamine transporter 2	VMAT2	N
Absent in melanoma 1 gene	AIM1	G
Acrosin	ACR	G
Activin		G
Activin A receptor, type 2-like kinase 1	ACVRL1	G
Activin A receptor, type 2B	ACVR2B	G
Adenomatous polyposis coli tumour supressor gene	APC	G
Adrenocorticotrophic hormone (ACTH) receptor	ACTHR	G
Aldosterone receptor	MLR	G
Alkaptonuria gene	AKU	G
alpha tectorin	TECTA	G
alpha-actinin 2	ACTN2	G
alpha-actinin 3	ACTN3	G
Alpha-fetoprotein	AFP	G
Amphiregulin	AREG	G
Androgen receptor	AR	G
Angiopoietin 1	ANGPT1	G
Angiopoietin 2	ANGPT2	G
Anti-Mullerian hormone	AMH	G
Anti-Mullerian hormone type 2 receptor	AMHR2	G
AP-2, alpha	TFAP2A	G
AP-2, beta	TFAP2B	G
AP-2, gamma	TFAP2C	G
Apical protein, xenopus laevis-like	APXL	G
Apopain	CPP32	G
Archaete-scute homolog 1	ASH1	G
Archaete-scute homolog 2	ASH2	G
Astrotactin	ASTN	G
Ataxia telangiectasia complementation group D	ATD, ATDC	G
Ataxia telangiectasia gene, AT	ATM	G
Ataxin 1	SCA1	G
Ataxin 2	SCA2	G
Ataxin 3	MJD	G
Atrial natriuretic peptide	ANP	G
Atrial natriuretic peptide receptor A	NPR1	G
Atrial natriuretic peptide receptor B	NPR2	G
Atrial natriuretic peptide receptor C	NPR3	G
Atrophin 1	DRPLA	G
Azoospermia factor 1	AZF1	G
Bagpipe homeobox, drosophila homolog of, 1	BAPX1	G
BCL2-associated X protein	BAX	G
BCL2-related protein A1	BCL2A1	G
Beckwith-Wiedemann region 1A	BWR1A	G
Bloom syndrome protein	BLM	G
Bone morphogenetic protein, BMP1	BMP1	G

Bone morphogenetic protein, BMP2	BMP2	G
Bone morphogenetic protein, BMP3	BMP3	G
Bone morphogenetic protein, BMP4	BMP4	G
Bone morphogenetic protein, BMP5	BMP5	G
Bone morphogenetic protein, BMP6	BMP6	G
Bone morphogenetic protein, BMP7	BMP7	G
Bone morphogenetic protein, BMP8	BMP8	G
Brain derived neurotrophic factor	BDNF	G
Brain derived neurotrophic factor (BDNF) receptor	BDNFR	G
BRCA1-associated RING domain gene 1	BARD1	G
Breakpoint cluster region	BCR	G
Breast cancer 1	BRCA1	G
Breast cancer 2	BRCA2	G
Breast cancer, ductal, 1	BRCD1	G
Breast cancer, ductal, 2	BRCD2	G
Bruton agammaglobulinaemia tyrosine kinase	BTK	G
Cadherin E	CDH1	G
Cadherin EP		G
Cadherin N	CDH2	G
Cadherin P	CDH3	G
Calbindin 1	CALB1	G
Calbindin D9K	CALB3	G
Calmodulin 1	CALM1	G
Calmodulin 2	CALM2	G
Calmodulin 3	CALM3	G
Calmodulin-dependant protein kinase II	CAMK2A	G
Calnexin	CANX	G
Cardiac-specific homeobox, CSX	CSX	G
Caspase 1	CASP1	G
Caspase 10	CASP10	G
Caspase 2	CASP2	G
Caspase 3	CASP3	G
Caspase 4	CASP4	G
Caspase 5	CASP5	G
Caspase 6	CASP6	G
Caspase 7	CASP7	G
Caspase 8	CASP8	G
Caspase 9	CASP9	G
Catenin, alpha	CTNNA1	G
Catenin, beta	CTNNB1	G
Catenin, gamma		G
Cdc 25 phosphatase		G
Cdc2	CDC2	G
CDX1		G
CEA		G
Cell adhesion molecule, intercellular, ICAM	ICAM1	G
Cell adhesion molecule, leukocyte-endothelial, LECAM (CD62)	LECAM1	G
Cell adhesion molecule, liver, LCAM	LCAM	G
Cell adhesion molecule, neural, NCAM1	NCAM1	G

Cell adhesion molecule, neural, NCAM120	NCAM120	G
Cell adhesion molecule, neural, NCAM2	NCAM2	G
Cell adhesion molecule, platelet-endothelial, PECAM	PECAM1	G
Cell adhesion molecule, vascular, VCAM	VCAM1	G
c-erbB1	ERBB1	G
c-erbB2	ERBB2	G
c-erbB3	ERBB3	G
c-erbB4	ERBB4	G
Cholestasis, progressive familial intrahepatic 1 gene	FIC1	G
Chromogranin A	CHGA	G
Ciliary neurotrophic factor (CNTF)	CNTF	G
Ciliary neurotrophic factor (CNTF) receptor	CNTFR	G
c-kit receptor tyrosine kinase		G
Cleavage signal-1 protein	CS1	G
Cleft palate gene	CPX	G
Clusterin	CLU	G
Cockayne syndrome gene, CKN1	CKN1	G
Collapsin		G
Colony-stimulating factor 1	CSF1	G
Colony-stimulating factor 1 receptor	CSF1R	G
Colony-stimulating factor 2	CSF2	G
Colony-stimulating factor 2 alpha receptor	CSF2RA	G
Colony-stimulating factor 2 beta receptor	CSF2RB	G
Colony-stimulating factor 3	CSF3	G
Colony-stimulating factor 3 receptor	CSF3R	G
Cone-rod homeobox-containing gene	CRX	G
Contactin	CNTN1	G
Core-binding factor, alpha 1	CBFA1	G
Core-binding factor, alpha 2	CBFA2	G
Core-binding factor, beta	CBFB	G
Creb binding protein	CREBBP	G
c-src tyrosine kinase	CSK	G
Cyclic AMP response element binding protein	CREB	G
Cyclic AMP response element modulator	CREM	G
Cyclic AMP-dependent protein kinase	PKA	E
Cyclin A	CCNA	G
Cyclin B	CCNB	G
Cyclin C	CCNC	G
Cyclin D	CCND1	G
Cyclin E	CCNE	G
Cyclin F	CCNF	G
Cyclin-dependent kinase 1	CDK1	G
Cyclin-dependent kinase 10	CDK10	G
Cyclin-dependent kinase 2	CDK2	G
Cyclin-dependent kinase 3	CDK3	G
Cyclin-dependent kinase 4	CDK4	G
Cyclin-dependent kinase 5	CDK5	G
Cyclin-dependent kinase 6	CDK6	G
Cyclin-dependent kinase 7	CDK7	G

Cyclin-dependent kinase 8	CDK8	G
Cyclin-dependent kinase 9	CDK9	G
Cyclin-dependent kinase inhibitor 1A (P21, CIP1)	CDKN1A	G
Cyclin-dependent kinase inhibitor 1B (P27, KIP1)	CDKN1B	G
Cyclin-dependent kinase inhibitor 1C (P57, KIP2)	CDKN1C	G
Cyclin-dependent kinase inhibitor 2A (p16)	CDKN2A	G
Cyclin-dependent kinase inhibitor 3	CDKN3	G
Defender against cell death 1	DAD1	G
Deleted in azoospermia	DAZ	G
Deleted in colorectal carcinoma	DCC	G
Deleted in malignant brain tumours 1	DMBT1	G
Dentin sialophosphoprotein	DSPP	G
Desert hedgehog, dhh		G
Disrupted meiotic cDNA 1, homolog	DMC1	G
Distal-less homeobox 1	DLX1	G
Distal-less homeobox 2	DLX2	G
Distal-less homeobox 3	DLX3	G
Distal-less homeobox 4	DLX4	G
Distal-less homeobox 5	DLX5	G
Distal-less homeobox 6	DLX6	G
Dynamin	DNM1	G
Dynein		G
E74-like factor 1, ELF1	ELF1	G
EB1		G
Empty spiracles (drosophila) homologue 1	EMX1	G
Empty spiracles (drosophila) homologue 2	EMX2	G
Endometrial bleeding-associated factor	EBAF	G
Engrailed-1	EN1	G
Engrailed-2	EN2	G
Ephrin receptor tyrosine kinase A	EPHA	G
Ephrin receptor tyrosine kinase B	EPHB	G
Ephrin-A	EFNA	G
Ephrin-B	EFNB	G
Epidermal growth factor	EGF	G
Epidermal growth factor receptor	EGFR	G
Erythroid kruppel-like factor	EKLF	G
Estrogen receptor	ESR	G
Eukaryotic initiation translation factor	EIF4E	G
EWS RNA-binding protein	EWSR1	G
Eyes absent 1	EYA1	G
Eyes absent 2	EYA2	G
Eyes absent 3	EYA3	G
Fc fragment of IgG, high affinity IA, receptor for	FCGR1A	G
Fc fragment of IgG, low affinity IIa, receptor for (CD32)	FCGR2A	G
Fc fragment of IgG, low affinity IIIa, receptor for (CD16)	FCGR3A	G
Fertilin protein	FTNB	G
Fibrillin 1	FBN1	G
Fibrillin 2	FBN2	G

Fibroblast growth factor	FGF1	G
Fibroblast growth factor receptor 1	FGFR1	G
Fibroblast growth factor receptor 2	FGFR2	G
Fibroblast growth factor receptor 3	FGFR3	G
Fibronectin precursor	FN1	G
Flightless-II, Drosophila homolog of	FLII	G
Folic acid receptor	FOLR	G
Follicle stimulating hormone receptor	FSHR, ODG1	G
Follicle stimulating hormone, FSH	FSHB	G
Follistatin		G
Forkhead rhabdomyosarcoma gene	FKHR	G
Forkhead transcription factor 10	FKHL10	G
Forkhead transcription factor 14	FKHL14	G
Forkhead transcription factor 7	FKHL7	G
Frataxin	FRDA	G
Fringe secreted protein, lunatic	LFNG	G
Fringe secreted protein, manic	MFNG	G
Fringe secreted protein, radical	RFNG	G
Fukuyama type congenital muscular dystrophy	FCMD	G
G/T mismatch binding protein	GTBP, MSH6	G
Galactosyltransferase 1	GT1	G
Galactosyltransferase, alpha 1,3	GGTA1	G
Galactosyltransferase, beta 3	B3GALT	G
Gastrin	GAS	G
Gastrulation brain homeobox 2	GBX2	G
GDP dissociation inhibitor 1	GDI1	G
Gelsolin	GSN	G
Geniospasm 1	GSM1	G
Glioma chloride ion channel, GCC		G
Glucagon receptor	GCGR	G
Glucagon-like peptide receptor 1	GLP1R	G
Glucocorticoid receptor	GRL	G
Glypican 3	GPC3, SDYS	G
Gonadotropin releasing hormone	GNRH	G
Gonadotropin releasing hormone receptor	GNRHR	G
Goosecoid GSC		G
Growth arrest-specific homeobox	GAX	G
Growth factor receptor-bound protein 2	GRB2	G
Growth hormone 1	GH1	G
Growth hormone 2 (placental)	GH2	G
Growth hormone receptor	GHR	G
Growth hormone releasing hormone (GHRH)	GHRH	G
Growth hormone releasing hormone receptor	GHRHR	G
Growth/differentiation factor 5	GDF5	G
GTP cylcohydrolase 1	GCH1	G
GTPase-activating protein, GAP	RASA1	G
Hairless	HR	G
Hela tumor suppression gene	HTS1	G
Heparin binding epidermal growth factor	HBEGF	G
Hepatocyte growth factor	HGF	G

High mobility group protein 1	HMG1	G
High mobility group protein 2	HMG2	G
High mobility group protein C	HMGIC	G
High mobility group protein Y	HMGIY	G
Histone family H1	H1	G
Histone family H2	H2	G
Histone family H3	H3	G
Histone family H4	H4	G
HLH transcription factor HAND1	HAND1	G
HLH transcription factor HAND2	HAND2	G
Holoprosencephaly 1	HPE1	G
Holoprosencephaly 2	HPE2	G
Holoprosencephaly 3	HPE3	G
Holoprosencephaly 4	HPE4	G
Homeobox (HOX) gene A1	HOXA1	G
Homeobox (HOX) gene A2	HOXA2	G
Homeobox (HOX) gene A3	HOXA3	G
Homeobox (HOX) gene A4	HOXA4	G
Homeobox (HOX) gene A5	HOXA5	G
Homeobox (HOX) gene A6	HOXA6	G
Homeobox (HOX) gene A7	HOXA7	G
Homeobox (HOX) gene A8	HOXA8	G
Homeobox (HOX) gene A9	HOXA9	G
Homeobox (HOX) gene A10	HOXA10	G
Homeobox (HOX) gene A11	HOXA11	G
Homeobox (HOX) gene A12	HOXA12	G
Homeobox (HOX) gene A13	HOXA13	G
Homeobox (HOX) gene B1	HOXB1	G
Homeobox (HOX) gene B2	HOXB2	G
Homeobox (HOX) gene B3	HOXB3	G
Homeobox (HOX) gene B4	HOXB4	G
Homeobox (HOX) gene B5	HOXB5	G
Homeobox (HOX) gene B6	HOXB6	G
Homeobox (HOX) gene B7	HOXB7	G
Homeobox (HOX) gene B8	HOXB8	G
Homeobox (HOX) gene B9	HOXB9	G
Homeobox (HOX) gene C4	HOXC4	G
Homeobox (HOX) gene C8	HOXC8	G
Homeobox (HOX) gene C9	HOXC9	G
Homeobox (HOX) gene C13	HOXC13	G
Homeobox (HOX) gene D1	HOXD1	G
Homeobox (HOX) gene D3	HOXD3	G
Homeobox (HOX) gene D4	HOXD4	G
Homeobox (HOX) gene D8	HOXD8	G
Homeobox (HOX) gene D9	HOXD9	G
Homeobox (HOX) gene D10	HOXD10	G
Homeobox (HOX) gene D12	HOXD12	G
Homeobox (HOX) gene D13	HOXD13	G
Homeobox 11	HOX11	G
Homeobox HB24	HLX1	G

Homeobox HB9	HLXB9	G
Homeobox, PROX1	PROX1	G
Human atonal gene	ATOH1	G
Human chorionic gonadtrophin, hCG	CG	G
Human placental lactogen	CSH1	G
Ikaros gene	IKAROS	G
Indian hedgehog, ihh	IHH	G
Inhibin, alpha	INHA	G
Inhibin, beta A	INHBA	G
Inhibin, beta B	INHBB	G
Inhibin, beta C	INHBC	G
Inositol 1,4,5-triphosphate receptor 1	ITPR1	G
Inositol 1,4,5-triphosphate receptor 3	ITPR3	G
Insulin	INS	G
Insulin promotor factor 1	IPF1	G
Insulin receptor	INSR	G
Insulin receptor substrate-1	IRS1	G
Insulin-like growth factor 1	IGF1	G
Insulin-like growth factor 1 receptor	IGF1R	G
Insulin-like growth factor 2	IGF2	G
Insulin-like growth factor 2 receptor	IGF2R	G
Integrin beta 1	ITGB1	G
Integrin beta 2	ITGB2	G
Integrin beta 3	ITGB3	G
Integrin beta 4	ITGB4	G
Integrin beta 5	ITGB5	G
Integrin beta 6	ITGB6	G
Integrin beta 7	ITGB7	G
Integrin, alpha 1	ITGA1	G
Integrin, alpha 2	ITGA2	G
Integrin, alpha 3	ITGA3	G
Integrin, alpha 4	ITGA4	G
Integrin, alpha 5	ITGA5	G
Integrin, alpha 6	ITGA6	G
Integrin, alpha 7	ITGA7	G
Integrin, alpha 8	ITGA8	G
Integrin, alpha 9	ITGA9	G
Integrin, alpha M	ITGAM	G
Integrin, alpha X	ITGAX	G
Janus kinase 1	JAK1	G
Janus kinase 2	JAK2	G
Janus kinase 3	JAK3	G
Kallman syndrome gene 1	KAL1	G
Kinectin	KTN1	G
Kinesin, heavy chain	KNSL1	G
Kinesin, light chain	KNS2	G
Lamin A/C	LMNA	G
Laminin 5, alpha 3	LAMA3	G
Laminin 5, beta 3	LAMB3	G
Laminin 5, gamma 2	LAMC2	G

Laminin M	LAMM	G
Laminin receptor 1	LAMR1	G
Latent transforming growth factor-beta binding protein 2	LTBP2	G
Leptin	LEP	G
Leptin receptor	LEPR	G
Leukaemia inhibitory factor	LIF	G
Leukaemia inhibitory factor receptor	LIFR	G
LH/choriogonadotropin (CG) receptor	LHCGR	G
LIM homeobox protein 1	LHX1	G
LIM homeobox protein 2	LHX2	G
LIM homeobox protein 3	LHX3	G
LIM homeobox protein 4	LHX4	G
LIM homeobox transcription factor 1, beta	LMX1B	G
Limb girdle muscular dystrophy 1A	LGMD1A	G
Limb girdle muscular dystrophy 1B	LGMD1B	G
Limb girdle muscular dystrophy 2G	LGMD2G	G
Limb girdle muscular dystrophy 2H	LGMD2H	G
Limbic associated membrane protein	LAMP	G
LIM-domain only protein 1	LMO1	G
LIM-domain only protein 2	LMO2	G
LIM-domain only protein 3	LMO3	G
LIM-domain only protein 4	LMO4	G
Lipoma-preferred partner gene	LPP	G
Luteinizing hormone, beta chain	LHB	G
Lymphoid enhancer-binding factor	LEF-1	G
Lysosome-associated membrane protein 1	LAMP1	G
Lysosome-associated membrane protein 2	LAMP2	G
MAD (mothers against decapentaplegic, Drosophila) homologue 2	MADH2	G
MAD (mothers against decapentaplegic, Drosophila) homologue 3	MADH3	G
MAD (mothers against decapentaplegic, Drosophila) homologue 4	MADH4	G
MADS box transcription-enhancer factor 2A	MEF2A	G
MADS box transcription-enhancer factor 2B	MEF2B	G
MADS box transcription-enhancer factor 2C	MEF2C	G
MADS box transcription-enhancer factor 2D	MEF2D	G
MAPK kinase 1	MAPKK1; MEK1	G
MAPK kinase 4	MAPKK4; MEK4; SERK1	G
MAPK kinase 6	MAPKK6; MEK6	G
MAPKK kinase	MAPKKK	G
Matrix Gla protein	MGP	G
MAX-interacting protein 1	MXI1	G
Menin	MEN1	G
Mesoderm-specific transcript	MEST	G
Microphthalmia-associated transcription factor	MITF	G
Midline 1	MID1	G
Mismatch repair gene, PMSL1	PMS1	G

Mismatch repair gene, PMSL2	PMS2	G
Mitogen-activated protein (MAP) kinase	MAPK	G
Motilin	MLN	G
Msh homeobox homolog 1	MSX1	G
Msh homeobox homolog 2	MSX2	G
Multidrug resistance associated protein	MRP	G
Mutated in colorectal cancers, MCC	MCC	G
MutL homolog 1	MLH1	G
MutS homolog 2	MSH2	G
MutS homolog 3	MSH3	G
Myelodysplasia syndrome 1 gene	MDS1	G
Myogenic factor 3	MYF3	G
Myogenic factor 4	MYF4	G
Myogenic factor 5	MYF5	G
Na+, K+ ATPase, alpha	ATP1A1	G
Na+, K+ ATPase, beta 1	ATP1B1	G
Na+, K+ ATPase, beta 2	ATP1B2	G
Na+, K+ ATPase, beta 3	ATP1B3	G
Necdin	NDN	G
Nerve growth factor	NGF	G
Nerve growth factor receptor	NGFR	G
Neural retina-specific gene	NRL	G
Neuregulin	HGL	G
Neurofibromin 1	NF1	G
Neurofibromin 2	NF2	G
Neurotrophic tyrosine kinase receptor 1	NTRK1	G
Neurotrophin 3	NTF3 or NT3	G
Neurturin	NRTN	G
Niacin receptor		G
Nibrin	NBS1	G
Nodal	NODAL	G
Noggin	NOG	G
Norrie disease protein	NDP	G
Notch 1	NOTCH1	G
Notch 2	NOTCH2	G
Notch 3	NOTCH3	G
Notch ligand - jagged 1	JAG1, AGS	G
Nuclear factor of activated T cells (NFAT) complex, cytosolic	NFATC	G
Nuclear factor of activated T cells (NFAT) complex, preexisting component	NFATP	G
Nuclear mitotic apparatus protein 1	NUMA1	G
Oligophrenin-1	OPHN1	G
Oncogene abl1	ABL1	G
Oncogene abl2		G
Oncogene akt1		G
Oncogene akt2	AKT2	G
Oncogene axl	AXL	G
Oncogene bcl2		G
Oncogene bcr/abl		G

Oncogene B-lym		G
Oncogene B-raf		G
Oncogene clk1		G
Oncogene c-myc		G
Oncogene cot		G
Oncogene crk		G
Oncogene crkl		G
Oncogene ect2		G
Oncogene ELK1	ELK1	G
Oncogene ELK2	ELK2	G
Oncogene ems1		G
Oncogene ERB		G
Oncogene ERB2		G
Oncogene ERBA		G
Oncogene ERBAL2		G
Oncogene ERG (early reponse gene)		G
Oncogene ETS1		G
Oncogene ETS2		G
Oncogene EVI1	EVI1	G
Oncogene fes		G
Oncogene fgr		G
Oncogene fos	FOS	G
Oncogene fps		G
Oncogene GLI1	GLI	G
Oncogene GLI2	GLI2	G
Oncogene GLI3	GLI3	G
Oncogene gro1		G
Oncogene gro2		G
Oncogene Ha-ras	HRAS	G
Oncogene hs1		G
Oncogene hst	FGF4	G
Oncogene int1	WNT1	G
Oncogene int2	FGF3	G
Oncogene int3	Notch4	G
Oncogene int4	WNT3	G
Oncogene jun	JUN	G
Oncogene KIT	KIT, PBT	G
Oncogene LCO	LCO	G
Oncogene l-myc		G
Oncogene lpsa		G
Oncogene lyn		G
Oncogene maf		G
Oncogene mas1		G
Oncogene mcf2		G
Oncogene mdm2	MDM2	G
Oncogene mel		G
Oncogene met	MET	G
Oncogene mos		G
Oncogene mpl		G
Oncogene MUM1	MUM1	G

Oncogene myb	MYB	G
Oncogene myc	MYC	G
Oncogene n-myc		G
Oncogene N-ras (neuroblastoma v-ras)	NRAS	G
Oncogene ovc		G
Oncogene pim1		G
Oncogene pti-1sea		G
Oncogene pvt1		G
Oncogene raf	RAF	G
Oncogene ralb		G
Oncogene rel		G
Oncogene ret	RET	G
Oncogene r-myc		G
Oncogene ros		G
Oncogene R-ras		G
Oncogene sis	PDGFB	G
Oncogene ski		G
Oncogene sno		G
Oncogene spi1		G
Oncogene src		G
Oncogene tc21		G
Oncogene TEL	ETV6	G
Oncogene tim		G
Oncogene vavtrk		G
Oncogene v-Ki-ras2	KRAS2	G
Oncogene yes		G
Oncogene yuasa		G
Oncostatin M	OSM	G
Oncostatin M receptor	OSMR	G
Orexin	OX	G
Orexin 1 receptor	OX1R	G
Orexin 2 receptor	OX2R	G
Orthodenticle (Drosophila) homolog 1	OTX1	G
Orthodenticle (Drosophila) homolog 2	OTX2	G
Osteonectin	ON	G
Osteopontin	OPN	G
Osteoprotegerin	OPG	G
p21-activated kinase 3	PAK3	G
Paired box homeotic gene 1	PAX1	G
Paired box homeotic gene 2	PAX2	G
Paired box homeotic gene 3	PAX3	G
Paired box homeotic gene 6	PAX6	G
Paired box homeotic gene 7	PAX7	G
Paired box homeotic gene 8	PAX8	G
Paired-like homeodomain transcription factor 2	PITX2	G
Paired-like homeodomain transcription factor 3	PITX3	G
Parathyroid hormone	PTH	G
Parathyroid hormone receptor	PTHR1	G
Parathyroid hormone related-peptide	PTHrP	G
Parathyroid hormone-like hormone	PTHLH	G

Parvalbumin	PVALB	G
Patched (Drosophila) homolog, PTCH	PTCH	G
Phosphatase & tensin homolog	PTEN	G
Phosphate regulating gene with homologies to endopeptidases on the X chromosome	PHEX	G
Phosphatidylinositol glycan, class A (paroxysmal nocturnal hemoglobinuria)	PIGA	G
Phosphatidylinositol transfer protein	PITPN	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 1	PDNP1	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 2	PDNP2	G
Phosphodiesterase 1 / nucleotide pyrophosphatase 3	PDNP3	G
Phosphomannomutase 1	PMM1	G
Phosphomannomutase 2	PMM2	G
Phytanoyl-CoA hydroxylase	PHYH	G
Platelet derived growth factor	PDGF	G
Platelet derived growth factor receptor	PDGFR	G
Poly(A) binding protein 2	PABP2	G
POU domain, class 1, transcription factor 1 (Pit1)	POU1F1	G
POU domain, class 3, transcription factor 4	POU3F4	G
POU domain, class 4, transcription factor 3	POU4F3	G
Pre-B-cell leukemia transcription factor 1	PBX1	G
Preproglucagon	GCG;GLP1; GLP2	G
Profibrinolysin		G
Progesterone receptor (RU486 binding receptor)	PGR	G
Prohibitin	PHB	G
Prolactin	PRL	G
Prolactin receptor	PRLR	G
Prolactin releasing hormone	PRH	G
Proliferin	PLF	G
Pro-melanin-concentrating hormone	PMCH	G
Promyelocytic leukemia gene	PML	G
Prophet of Pit1	PROP1	G
Prostaglandin (PG) D synthase, hematopoietic	PGDS	E
Prostaglandin isomerase		G
Prostaglandin-endoperoxidase synthase 2	PTGS2	G
Prostate cancer anti-metastasis gene KAI1	KAI1	G
Protein tyrosine phosphatase, non-receptor type 12	PTPN12	G
RAD51, DNA repair protein	RAD51	G
RAD52, DNA repair protein	RAD52	G
RAD54, DNA repair protein	RAD54	G
RAD55, DNA repair protein	RAD55	G
RAD57, DNA repair protein	RAD57	G
Ras-G-protein	RAS	G
Rathke pouch homeobox, RPX	RPX	G
Receptor tyrosine kinase (RTK), Nsk2	NSK2	G
Recombination activating gene 1	RAG1	G
Recombination activating gene 2	RAG2	G
Relaxin H1	RLN1	G
Relaxin H2	RLN2	G
Retinoblastoma 1	RB1	G

Retinoic acid receptor, alpha	RARA	G
Retinoic acid receptor, beta	RARB	G
Retinoic acid receptor, gamma	RARG	G
Retinoid X receptor, alpha	RXRA	G
Retinoid X receptor, beta	RXRB	G
Retinoid X receptor, gamma	RXRG	G
Retinoschisis, X-linked, juvenile	RS	G
Rhabdoid tumors	SMARCB1	G
RIGUI	RIGUI	G
Ryanodine receptor 1, skeletal	RYR1	G
SA homolog	SAH	G
Sal-like 1	SALL1	G
Serine/threonine kinase 11	STK11	G
Serine/threonine kinase 2	STK2	G
Sex determining region Y, SRY	SRY	G
Short stature homeobox	SHOX	G
Sialoprotein, bone	BSP	G
Signal transducer and activator of transcription 1	STAT1	G
Signal transducer and activator of transcription 2	STAT2	G
Signal transducer and activator of transcription 3	STAT3	G
Signal transducer and activator of transcription 4	STAT4	G
Signal transducer and activator of transcription 5	STAT5	G
Sine oculis homeobox, drosophila, homolog 1	SIX1	G
Sine oculis homeobox, drosophila, homolog 2	SIX2	G
Sine oculis homeobox, drosophila, homolog 5	SIX5	G
Slug protein		G
Smoothelin	SMTN	G
Smoothened (Drosophila) homolog	SMOH	G
Somatotrophin		G
Sonic hedgehog, SHH	SHH	G
SOS1 guanine nucleotide exchange factor	SOS1	G
Spastic paraplegia 7	SPG7	G
Sperm adhesion molecule	SPAM1	G
Sperm protamine P1	PRM1	G
Sperm protamine P2	PRM2	G
Split hand/foot malformation gene	DSS1	G
SRY-box 10	SOX10	G
SRY-box 11	SOX11	G
SRY-box 3	SOX3	G
SRY-box 4	SOX4	G
SRY-box 9	SOX9	G
Stem cell factor	SCF	G
Steroid hormone receptor responsive DNA elements		G
Stromal derived factor 1	SDF1	G
Sulfamidase	SGSH	G
Sulfonylurea receptor	SUR	G
Suppression of tumorigenicity 3 gene	ST3	G
Suppression of tumorigenicity 8 gene	ST8	G
Surfeit 1	SURF1	G
Syndecan 1	SYND1	G

Syndecan 2	SYND2	G
Syndecan 3	SYND3	G
Syndecan 4	SYND4	G
Synovial sarcoma gene 1	SSX1	G
Synovial sarcoma gene 2	SSX2	G
Talin	TLN	G
TATA binding protein	TBP	G
TATA binding protein associated factor 2A	TAF2A	G
TATA binding protein associated factor 2C2	TAF2C2	G
TATA binding protein associated factor 2D	TAF2E	G
TATA binding protein associated factor 2F	TAF2F	G
TATA binding protein associated factor 2H	TAF2H	G
TATA binding protein associated factor 2I	TAF2I	G
TATA binding protein associated factor 2J	TAF2J	G
TATA binding protein associated factor 2K	TAF2K	G
T-BOX 1	TBX1	G
T-BOX 2	TBX2	G
T-BOX 3	TBX3	G
T-BOX 4	TBX4	G
T-BOX 5	TBX5	G
T-BOX 6	TBX6	G
Testis-specific protein Y	TSPY	G
Thrombopoietin	THPO	G
Thrombospondin	THBS1	G
Thymopoietin	TMPO	G
Thyroglobulin	TG	G
Thyroid hormone receptor, alpha	THRA	G
Thyroid hormone receptor, beta	THRB	G
Thyroid peroxidase	TPO	G
Thyroid receptor auxiliary protein	TRAP	G
Thyroid-stimulating hormone receptor	TSHR	G
Thyroid-stimulating hormone, alpha	TSHA	G
Thyroid-stimulating hormone, beta	TSHB	G
Thyrotroph embryonic factor	TEF	G
Thyrotropin releasing hormone	TRH	G
Thyrotropin releasing hormone receptor	TRHR	G
TIE receptor tyrosine kinase	TIE-1	G
Torticollis, keloids, cryptorchidism and renal dysplasia gene	TKCR	G
Transcription factor 1, hepatic	TCF1	G
Transcription factor 2, hepatic	TCF2	G
Transcription factor 3	TCF3	G
Transcription factor binding to IGHM enhancer 3	TFE3	G
Transcription termination factor, RNA polymerase 1	TTF1	G
Transcription termination factor, RNA polymerase 2	TTF2	G
Transcription termination factor, RNA polymerase 3	TTF3	G
Transferrin	TF	G

Transferrin receptor	TFRC	G
Transforming growth factor, alpha	TGFA	G
Transforming growth factor, beta 2	TGFB2	G
Transforming growth factor, beta induced	TGFBI	G
Transforming growth factor, beta receptor 2	TGFBR2	G
Transglutaminase 1	TGM1	G
Transglutaminase 2	TGM2	G
Transglutaminase 4	TGM4	G
Translocation in renal carcinoma on chromosome 8 gene	TRC8	G
Treacle gene	TCOF1	G
Tubby-like protein 1	TULP1	G
Tuberous sclerosis 1	TSC1	G
Tuberous sclerosis 2	TSC2	G
Tumor susceptibility gene 101	TSG101	G
Tumour protein p53	TP53, P53	G
Tumour protein p63	TP63	G
Tumour protein p73	TP73	G
Tumour protein, translationally-controlled 1	TPT1	G
Twist (Drosophila) homolog	TWIST	G
Ubiquitin		G
Ubiquitin B	UBB	G
Ubiquitin C	UBC	G
Ubiquitin carboxyl-terminal esterase L1	UCHL1	G
Ubiquitin fusion degeneration 1-like	UFD1L	G
Vascular endothelial growth factor	VEGF	G
Vasoinhibitory peptide		G
Vitamin B12-binding (R) protein		G
Vitamin D receptor	VDR	G
v-myc avian myelocytomatosis viral oncogene homolog	MYC	G
Von Hippel-Lindau gene	VHL	G
Werner syndrome helicase	WRN	G
Wilms tumour gene 1	WT1	G
Wilms tumour gene 2	WT2	G
Wilms tumour gene 4	WT4	G
Winged helix nude	WHN	G
Wingless family, wnt2	WNT2	G
Wingless family, wnt4	WNT4	G
Wingless family, wnt5	WNT5	G
Wingless family, wnt7	WNT7	G
Wingless family, wnt8	WNT8	G
Wnt inhibitory factor, WIF-1	WIF1	G
Wolf-Hirschhorn syndrome candidate 1 gene	WHSC1	G
X (inactive)-specific transcript	XIST	G
X-ray repair gene	XRCC9	G
YY1 transcription factor	YY1	G
Zona pellucida glycoprotein 1	ZP1	G
Zona pellucida glycoprotein 2	ZP2	G
Zona pellucida glycoprotein 3	ZP3	G

Zona pellucida receptor tyrosine kinase	ZRK	G
Zonadhesin	ZAN	G

2. A set of probes, said probes being antibodies or antibody fragments which interact with specific expressed proteins encoded by gene sequences of a group of genes, said probes being for detecting relevant variants (mutations and polymorphisms), e.g. nucleotide substitutions (missense, nonsense, splicing and regulatory), small deletions, small insertions, small insertion deletions, gross insertions, gross deletions, duplications, complex rearrangements and repeat variations in a target group of genes; characterised in that said group is a core group of genes consisting of substantially all of the genes defined in claim 1.

3. A set according to claim 1 or 2 in which a minority of said probes for listed genes are absent.

4. A set according to claim 1 or 2 in which a limited number of additional probes are present together with substantially all of the probes for the listed genes.

5. A set according to claim 1 or 2 in which a limited number of probes are replaced by probes for non-listed genes.

6. A set of probes for a core group of genes according to any of claims 1 to 5 in which each gene to be probed is substantially similar (greater than 85% homologous) in sequence to the respective member of the core list of genes.

7. A set according to any of claims 1 to 6 consisting of probes for members of a sub-group of the core group.

8. A set according to any preceding claim in which said probes are in the form of an array and are spatially arranged at known locations on a substrate.

9. A set according to any preceding claim wherein said probes are on a substrate which forms part of or consists of one or more chip plate(s), for use in a chip assay for detection of said gene variants.

10. A set according to any preceding claim in which said probes are mass, electrostatic or fluorescence tagged probes.

11. A set according to claim 8 or 9 in which said substrate is a semiconductor microchip.

12. A set according to any preceding claim for use in a biological assay for detection of said gene variants.

13. A set according to any preceding claim for use in the measurement of differential gene expression levels.

14. A medical device including a set according to any preceding claim for use in an assay for detection of said gene variants.

15. A medical device including a set according to any of claims 1 to 13 for use in an array for detection of differential gene expression levels.

16. A method for use in assessing the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 1) in a target group of genes by hybridising a nucleic acid-containing sample from said patient or individual to a set according to any of claims 1 and 3 to 13 and relating the probe hybridisation pattern to said variations.

17. A method for use in assessing the the genomic profile of a patient or individual, the method comprising testing for and detecting the presence or absence of DNA or RNA encoding the relevant structural variants (as defined in claim 2) in a target group of genes by interacting an expressed-protein-containing sample from said patient or individual with a set of probes according to any of claims 2 to 13 and relating the probe interaction pattern to said variations.
18. Use of a set or device according to any of claims 1 to 13 for the prognosis and management of patients suffering from or at risk of disease.
19. Use of a set or device according to any of claims 1 to 13 for predicting likely therapeutic response and adverse events following therapeutic intervention.
20. Use of a set or device according to any of claims 1 to 13 for predicting likely patterns of symptom clusters (symptom profiles) in disease and the likelihood of subsequent, contingent, disease or symptoms.
21. Use of a set or device according to any of claims 1 to 13 for general health screening, occupational health purposes, healthcare planning on a population basis and other healthcare management utilisations.
22. Use of a set or device according to any of claims 1 to 13 for the development of new strategies of therapeutic intervention and in clinical trials.
23. Use of a set or device according to any of claims 1 to 13 for construction of and generation of algorithms for patient and healthcare management.
24. Use of a set or device according to any of claims 1 to 13 for modelling or assessing the impact of diseases or healthcare management strategies on individuals, groups, patient cohorts or populations
25. Use of a set or device according to any of claims 1 to 13 for modelling, assessing or exploring the theoretical impact of diseases and healthcare management strategies on individuals, groups, patient cohorts or populations.
26. Use of a set or device according to any of claims 1 to 13 for predicting optimum configuration/management of thereapeutic intervention.
27. A method according to claim 16 or 17 in which the identification of gene variants is indicative of a higher risk of developing clinical symptoms for the patient or individual.
28. A method for generating a model to assess whether a patient or individual or population or group is or are likely to develop clinical symptoms which method comprises:

i) obtaining DNA or RNA or protein samples from patients or individuals diagnosed as suffering from symptoms;
ii) obtaining DNA or RNA or protein samples from a control group of subjects diagnosed as not suffering from the symptoms;
iii) analysing the samples obtained in i) and ii) to identify the polymorphic variations encoded in the core group of genes as defined in any of claims 1 to 7;
iv) calculating the frequencies of these alleles in the samples from i) and ii);
v) comparing the frequencies of these alleles in i) and ii);
vi) performing a statistical analysis on the results from v) in order to generate a model for assessing the risk of developing symptoms.

29. A method for assessing whether a given subject will be at risk of developing symptoms, which comprises comparing said subject's genotype with a model generated by the method of claim 28.

30. A method according to any of claims 16, 17, 28 and 29 wherein at least one step is computer-controlled.

31. An assay suitable for use in a method according to any of claims 16, 17, 28 and 29; said assay comprising means for determining the presence or absence of relevant polymorphic variants of the core group of genes as defined in any of claims 1 to 7 in a biological sample.

32. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing symptoms; said kit comprising:

i) means for testing for the presence or absence or DNA or RNA encoding relevant polymorphic variants of the core group of genes as defined in claim 1 or 3 to 7 in a sample of human DNA;

ii) reagents for use in the detection process

iii) readout indicating the probability of a patient or individual developing symptoms.

33. A formatted assay technique (kit) for use in assessing the risk of a patient or individual developing symptoms; said kit comprising:

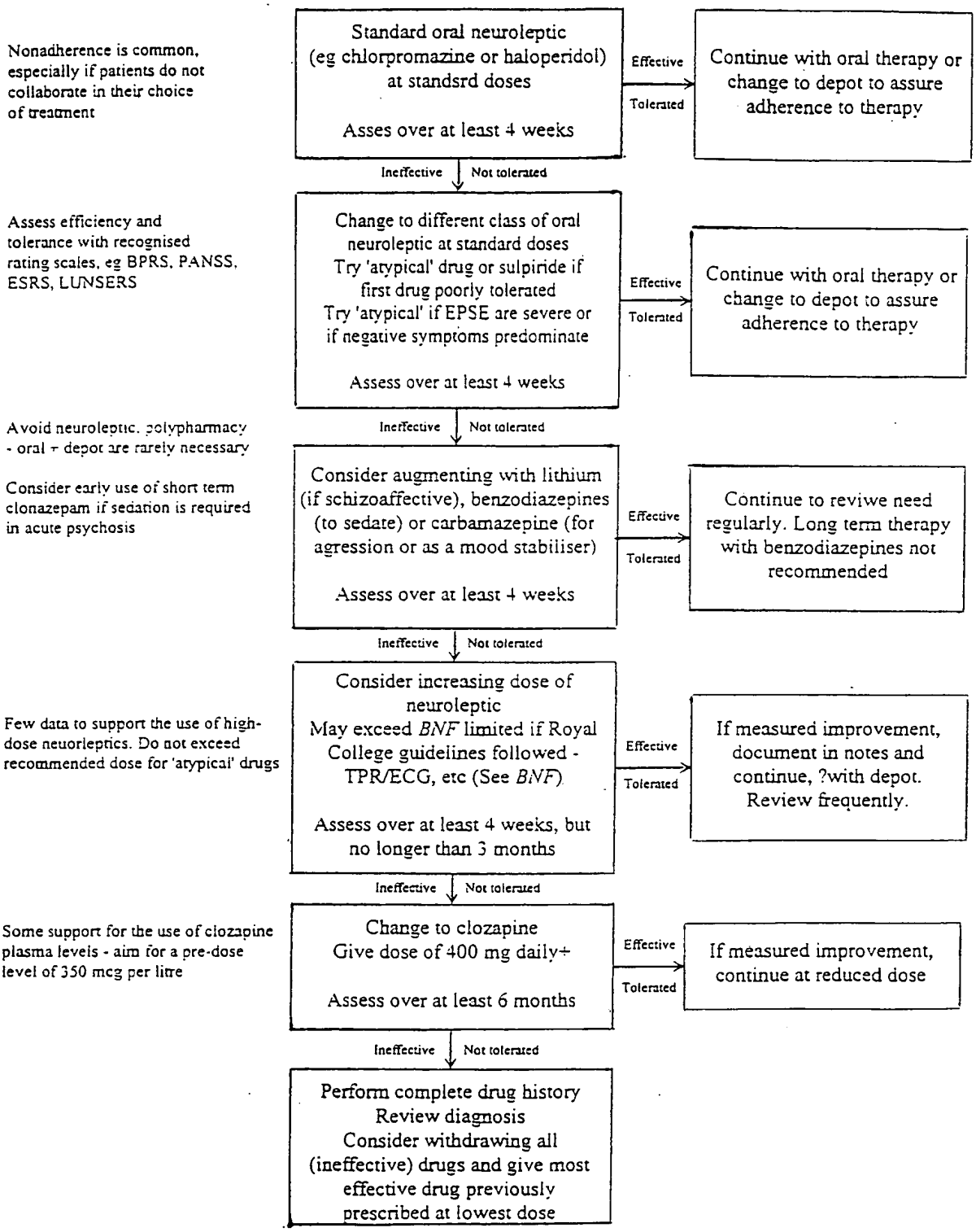
i) means for testing for the presence or absence of proteins encoded by the core group of genes and/or relevant polymorphic variants of the core group of genes as defined in any of claims 2 to 7 in an expressed-protein-containing human sample;

ii) reagents for use in the detection process

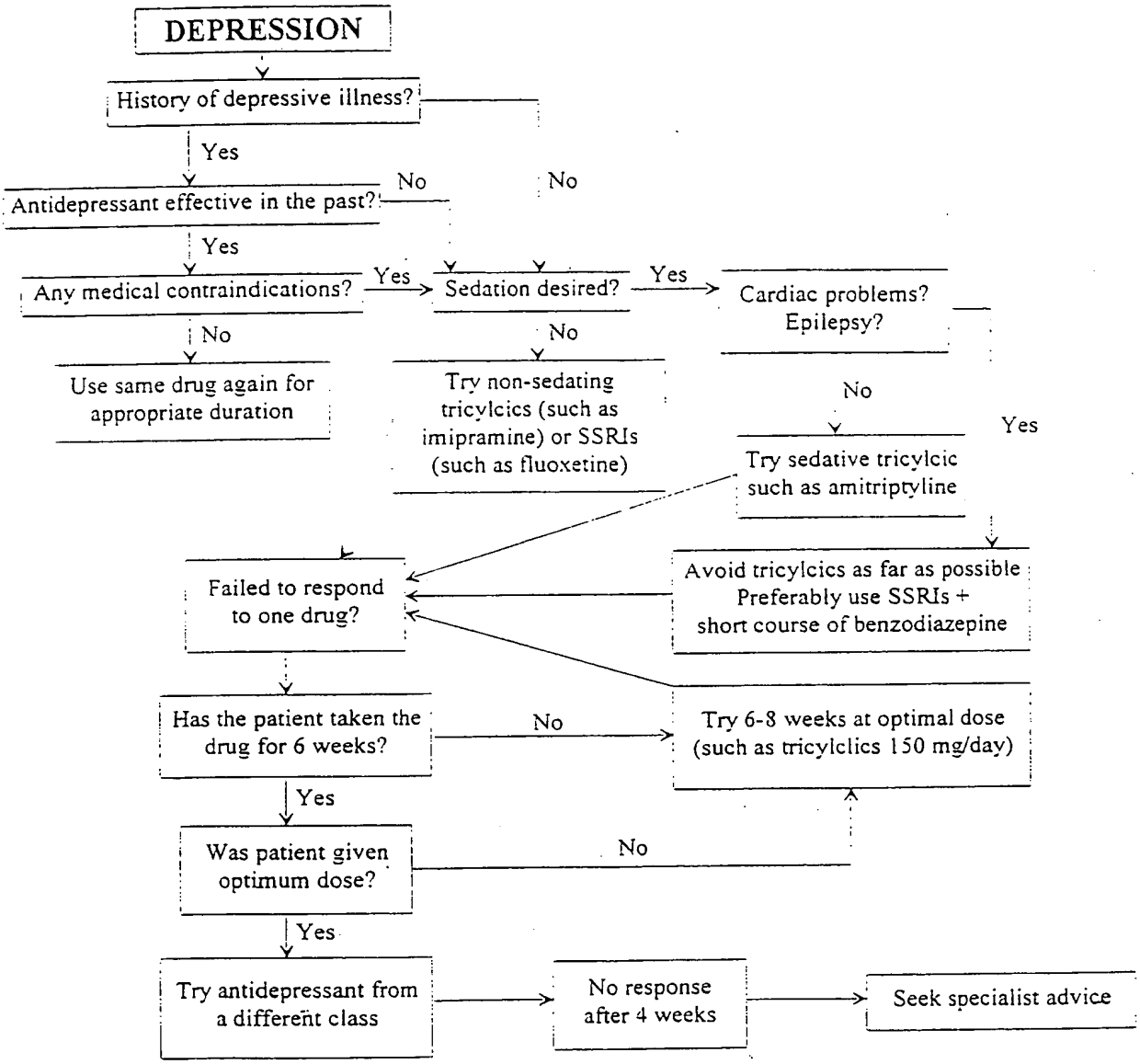
iii) readout indicating the probability of a patient or individual developing symptoms.

34. A set of probes according to claim 1, wherein the probes are selected from the group consisting of oligonucleotides and polynucleotides.

SCHIZOPHRENIA



DEPRESSION


DEPRESSION
History of depressive illness?
Yes
No
Antidepressant effective in the past?
No
Yes
Any medical contraindications?
Yes
Sedation desired?
Yes
Cardiac problems? Epilepsy?
No
Use same drug again for appropriate duration
No
Try non-sedating tricyclcics (such as imipramine) or SSRIs (such as fluoxetine)
No
Yes
Try sedative tricyclcic such as amitriptyline
Failed to respond to one drug?
Avoid tricyclcics as far as possible Preferably use SSRIs + short course of benzodiazepine
Has the patient taken the drug for 6 weeks?
No
Try 6-8 weeks at optimal dose (such as tricyclclics 150 mg/day)
Yes
Was patient given optimum dose?
No
Yes
Try antidepressant from a different class
No response after 4 weeks
Seek specialist advice